**Reference List for Microdeletion and Microduplication Syndromes**

1. Desai R, Frazier AE, Durigon R, Patel H, Jones AW, Dalla Rosa I, Lake NJ, Compton AG, Mountford HS, Tucker EJ *et al*: **ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism**. *Brain* 2017, **140**(6):1595-1610 [doi.org/10.1093/brain/awx094.](https://doi.org/10.1093/brain/awx094.)

2. Gunning AC, Strucinska K, Muñoz Oreja M, Parrish A, Caswell R, Stals KL, Durigon R, Durlacher-Betzer K, Cunningham MH, Grochowski CM *et al*: **Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism**. *American journal of human genetics* 2020, **106**(2):272-279 [doi.org/10.1016/j.ajhg.2020.01.007.](https://doi.org/10.1016/j.ajhg.2020.01.007.)

3. Wilson BT, Omer M, Hellens SW, Zwolinski SA, Yates LM, Lynch SA: **Microdeletion 1p35.2: a recognizable facial phenotype with developmental delay**. *American journal of medical genetics Part A* 2015, **167a**(8):1916-1920 [doi.org/10.1002/ajmg.a.37114.](https://doi.org/10.1002/ajmg.a.37114.)

4. Gennarino VA, Palmer EE, McDonell LM, Wang L, Adamski CJ, Koire A, See L, Chen C-A, Schaaf CP, Rosenfeld JA *et al*: **A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures**. *Cell* 2018, **172**(5):924-936.e911 [doi.org/10.1016/j.cell.2018.02.006.](https://doi.org/10.1016/j.cell.2018.02.006.)

5. Lu W, Quintero-Rivera F, Fan Y, Alkuraya FS, Donovan DJ, Xi Q, Turbe-Doan A, Li Q-G, Campbell CG, Shanske AL *et al*: **NFIA Haploinsufficiency Is Associated with a CNS Malformation Syndrome and Urinary Tract Defects**. *PLOS Genetics* 2007, **3**(5):e80 [doi.org/10.1371/journal.pgen.0030080.](https://doi.org/10.1371/journal.pgen.0030080.)

6. Koehler U, Holinski-Feder E, Ertl-Wagner B, Kunz J, von Moers A, von Voss H, Schell-Apacik C: **A novel 1p31.3p32.2 deletion involving the NFIA gene detected by array CGH in a patient with macrocephaly and hypoplasia of the corpus callosum**. *European journal of pediatrics* 2010, **169**(4):463-468 [doi.org/10.1007/s00431-009-1057-2.](https://doi.org/10.1007/s00431-009-1057-2.)

7. Chen C-P, Su Y-N, Chen Y-Y, Chern S-R, Liu Y-P, Wu P-C, Lee C-C, Chen Y-T, Wang W: **Chromosome 1p32-p31 deletion syndrome: Prenatal diagnosis by array comparative genomic hybridization using uncultured amniocytes and association with NFIA haploinsufficiency, ventriculomegaly, corpus callosum hypogenesis, abnormal external genitalia, and intrauterine growth restriction**. *Taiwanese Journal of Obstetrics and Gynecology* 2011, **50**(3):345-352 [doi.org/doi.org/10.1016/j.tjog.2011.07.014.](https://doi.org/https://doi.org/10.1016/j.tjog.2011.07.014.)

8. Ji J, Salamon N, Quintero-Rivera F: **Microdeletion of 1p32-p31 involving NFIA in a patient with hypoplastic corpus callosum, ventriculomegaly, seizures and urinary tract defects**. *European journal of medical genetics* 2014, **57**(6):267-268 [doi.org/10.1016/j.ejmg.2014.03.004.](https://doi.org/10.1016/j.ejmg.2014.03.004.)

9. Rao A, O'Donnell S, Bain N, Meldrum C, Shorter D, Goel H: **An intragenic deletion of the NFIA gene in a patient with a hypoplastic corpus callosum, craniofacial abnormalities and urinary tract defects**. *European journal of medical genetics* 2014, **57**(2-3):65-70 [doi.org/10.1016/j.ejmg.2013.12.011.](https://doi.org/10.1016/j.ejmg.2013.12.011.)

10. Kehrer M, Schäferhoff K, Bonin M, Jauch A, Bevot A, Tzschach A: **Interstitial 1p32.1p32.3 deletion in a patient with multiple congenital anomalies**. *American journal of medical genetics Part A* 2015, **167a**(10):2406-2410 [doi.org/10.1002/ajmg.a.37178.](https://doi.org/10.1002/ajmg.a.37178.)

11. Labonne JD, Shen Y, Kong IK, Diamond MP, Layman LC, Kim HG: **Comparative deletion mapping at 1p31.3-p32.2 implies NFIA responsible for intellectual disability coupled with macrocephaly and the presence of several other genes for syndromic intellectual disability**. *Molecular cytogenetics* 2016, **9**:24 [doi.org/10.1186/s13039-016-0234-z.](https://doi.org/10.1186/s13039-016-0234-z.)

12. Bayat A, Kirchhoff M, Madsen CG, Roos L, Kreiborg S: **Familial craniofacial abnormality and polymicrogyria associated with a microdeletion affecting the NFIA gene**. *Clinical dysmorphology* 2017, **26**(3):148-153 [doi.org/10.1097/mcd.0000000000000182.](https://doi.org/10.1097/mcd.0000000000000182.)

13. Carter MT, Nikkel SM, Fernandez BA, Marshall CR, Noor A, Lionel AC, Prasad A, Pinto D, Joseph-George AM, Noakes C *et al*: **Hemizygous deletions on chromosome 1p21.3 involving the DPYD gene in individuals with autism spectrum disorder**. *Clinical Genetics* 2011, **80**(5):435-443 [doi.org/doi.org/10.1111/j.1399-0004.2010.01578.x.](https://doi.org/https://doi.org/10.1111/j.1399-0004.2010.01578.x.)

14. Willemsen MH, Vallès A, Kirkels LA, Mastebroek M, Olde Loohuis N, Kos A, Wissink-Lindhout WM, de Brouwer AP, Nillesen WM, Pfundt R *et al*: **Chromosome 1p21.3 microdeletions comprising DPYD and MIR137 are associated with intellectual disability**. *Journal of medical genetics* 2011, **48**(12):810-818 [doi.org/10.1136/jmedgenet-2011-100294.](https://doi.org/10.1136/jmedgenet-2011-100294.)

15. D’Angelo CS, Varela MC, de Castro CIE, Otto PA, Perez ABA, Lourenço CM, Kim CA, Bertola DR, Kok F, Garcia-Alonso L *et al*: **Chromosomal microarray analysis in the genetic evaluation of 279 patients with syndromic obesity**. *Molecular cytogenetics* 2018, **11**(1):14 [doi.org/10.1186/s13039-018-0363-7.](https://doi.org/10.1186/s13039-018-0363-7.)

16. Klopocki E, Schulze H, Strauss G, Ott C-E, Hall J, Trotier F, Fleischhauer S, Greenhalgh L, Newbury-Ecob RA, Neumann LM *et al*: **Complex inheritance pattern resembling autosomal recessive inheritance involving a microdeletion in thrombocytopenia-absent radius syndrome**. *American journal of human genetics* 2007, **80**(2):232-240 [doi.org/10.1086/510919.](https://doi.org/10.1086/510919.)

17. Brunetti-Pierri N, Berg JS, Scaglia F, Belmont J, Bacino CA, Sahoo T, Lalani SR, Graham B, Lee B, Shinawi M *et al*: **Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities**. *Nature genetics* 2008, **40**(12):1466-1471 [doi.org/10.1038/ng.279.](https://doi.org/10.1038/ng.279.)

18. Mefford HC, Sharp AJ, Baker C, Itsara A, Jiang Z, Buysse K, Huang S, Maloney VK, Crolla JA, Baralle D *et al*: **Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes**. *New England Journal of Medicine* 2008, **359**(16):1685-1699 [doi.org/10.1056/NEJMoa0805384.](https://doi.org/10.1056/NEJMoa0805384.)

19. Rosenfeld JA, Traylor RN, Schaefer GB, McPherson EW, Ballif BC, Klopocki E, Mundlos S, Shaffer LG, Aylsworth AS, q21.1 Study G: **Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes**. *Eur J Hum Genet* 2012, **20**(7):754-761 [doi.org/10.1038/ejhg.2012.6.](https://doi.org/10.1038/ejhg.2012.6.)

20. Pang H, Yu X, Kim YM, Wang X, Jinkins JK, Yin J, Li S, Gu H: **Disorders Associated With Diverse, Recurrent Deletions and Duplications at 1q21.1**. *Frontiers in Genetics* 2020, **11**(577) [doi.org/10.3389/fgene.2020.00577.](https://doi.org/10.3389/fgene.2020.00577.)

21. Burkardt DD, Rosenfeld JA, Helgeson ML, Angle B, Banks V, Smith WE, Gripp KW, Moline J, Moran RT, Niyazov DM *et al*: **Distinctive phenotype in 9 patients with deletion of chromosome 1q24-q25**. *American journal of medical genetics Part A* 2011, **155a**(6):1336-1351 [doi.org/10.1002/ajmg.a.34049.](https://doi.org/10.1002/ajmg.a.34049.)

22. Chatron N, Haddad V, Andrieux J, Désir J, Boute O, Dieux A, Baumann C, Drunat S, Gérard M, Bonnet C *et al*: **Refinement of genotype-phenotype correlation in 18 patients carrying a 1q24q25 deletion**. *American journal of medical genetics Part A* 2015, **167a**(5):1008-1017 [doi.org/10.1002/ajmg.a.36856.](https://doi.org/10.1002/ajmg.a.36856.)

23. Lam F, Morris C: **Nine year old boy with chromosome 1q23.3-q25.1 deletion**. *American Journal of Medical Genetics Part A* 2016, **170**(11):3013-3017 [doi.org/doi.org/10.1002/ajmg.a.37843.](https://doi.org/https://doi.org/10.1002/ajmg.a.37843.)

24. Mackenroth L, Hackmann K, Klink B, Weber JS, Mayer B, Schröck E, Tzschach A: **Interstitial 1q23.3q24.1 deletion in a patient with renal malformation, congenital heart disease, and mild intellectual disability**. *American journal of medical genetics Part A* 2016, **170**(9):2394-2399 [doi.org/10.1002/ajmg.a.37785.](https://doi.org/10.1002/ajmg.a.37785.)

25. Fichera M, Saccuzzo L, Bertuzzo S, Marelli S, Cavallini A, Romaniello R, Kocova M, Citterio A, Fanizza I, Trabacca A *et al*: **Assigning single clinical features to their disease-locus in large deletions: the example of chromosome 1q23-25 deletion syndrome**. *Journal of Translational Genetics and Genomics* 2020, **4**(3):114-132 [doi.org/10.20517/jtgg.2020.16.](https://doi.org/10.20517/jtgg.2020.16.)

26. Rosenfeld JA, Lacassie Y, El-Khechen D, Escobar LF, Reggin J, Heuer C, Chen E, Jenkins LS, Collins AT, Zinner S *et al*: **New cases and refinement of the critical region in the 1q41q42 microdeletion syndrome**. *European journal of medical genetics* 2011, **54**(1):42-49 [doi.org/10.1016/j.ejmg.2010.10.002.](https://doi.org/10.1016/j.ejmg.2010.10.002.)

27. Au PY, Argiropoulos B, Parboosingh JS, Micheil Innes A: **Refinement of the critical region of 1q41q42 microdeletion syndrome identifies FBXO28 as a candidate causative gene for intellectual disability and seizures**. *American journal of medical genetics Part A* 2014, **164a**(2):441-448 [doi.org/10.1002/ajmg.a.36320.](https://doi.org/10.1002/ajmg.a.36320.)

28. Spreiz A, Haberlandt E, Baumann M, Baumgartner Sigl S, Fauth C, Gautsch K, Karall D, Janetschek C, Rostasy K, Scholl-Bürgi S *et al*: **Chromosomal microaberrations in patients with epilepsy, intellectual disability, and congenital anomalies**. *Clinical Genetics* 2014, **86**(4):361-366 [doi.org/doi.org/10.1111/cge.12288.](https://doi.org/https://doi.org/10.1111/cge.12288.)

29. Cassina M, Rigon C, Casarin A, Vicenzi V, Salviati L, Clementi M: **FBXO28 is a critical gene of the 1q41q42 microdeletion syndrome**. *American journal of medical genetics Part A* 2015, **167**(6):1418-1420 [doi.org/10.1002/ajmg.a.37033.](https://doi.org/10.1002/ajmg.a.37033.)

30. Yanagishita T, Yamamoto-Shimojima K, Nakano S, Sasaki T, Shigematsu H, Imai K, Yamamoto T: **Phenotypic features of 1q41q42 microdeletion including WDR26 and FBXO28 are clinically recognizable: The first case from Japan**. *Brain & development* 2019, **41**(5):452-455 [doi.org/10.1016/j.braindev.2018.12.006.](https://doi.org/10.1016/j.braindev.2018.12.006.)

31. Ballif BC, Rosenfeld JA, Traylor R, Theisen A, Bader PI, Ladda RL, Sell SL, Steinraths M, Surti U, McGuire M *et al*: **High-resolution array CGH defines critical regions and candidate genes for microcephaly, abnormalities of the corpus callosum, and seizure phenotypes in patients with microdeletions of 1q43q44**. *Human genetics* 2012, **131**(1):145-156 [doi.org/10.1007/s00439-011-1073-y.](https://doi.org/10.1007/s00439-011-1073-y.)

32. Thierry G, Bénéteau C, Pichon O, Flori E, Isidor B, Popelard F, Delrue M-A, Duboscq-Bidot L, Thuresson A-C, van Bon BWM *et al*: **Molecular characterization of 1q44 microdeletion in 11 patients reveals three candidate genes for intellectual disability and seizures**. *American Journal of Medical Genetics Part A* 2012, **158A**(7):1633-1640 [doi.org/doi.org/10.1002/ajmg.a.35423.](https://doi.org/https://doi.org/10.1002/ajmg.a.35423.)

33. Depienne C, Nava C, Keren B, Heide S, Rastetter A, Passemard S, Chantot-Bastaraud S, Moutard ML, Agrawal PB, VanNoy G *et al*: **Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU**. *Human genetics* 2017, **136**(4):463-479 [doi.org/10.1007/s00439-017-1772-0.](https://doi.org/10.1007/s00439-017-1772-0.)

34. Chen CP, Ko TM, Wang LK, Chern SR, Wu PS, Chen SW, Wu FT, Chen YY, Chen WL, Wang W: **Prenatal diagnosis and molecular cytogenetic characterization of a chromosome 1q42.3-q44 deletion in a fetus associated with ventriculomegaly on prenatal ultrasound**. *Taiwanese journal of obstetrics & gynecology* 2020, **59**(4):598-603 [doi.org/10.1016/j.tjog.2020.05.022.](https://doi.org/10.1016/j.tjog.2020.05.022.)

35. Doco-Fenzy M, Leroy C, Schneider A, Petit F, Delrue M-A, Andrieux J, Perrin-Sabourin L, Landais E, Aboura A, Puechberty J *et al*: **Early-onset obesity and paternal 2pter deletion encompassing the ACP1, TMEM18, and MYT1L genes**. *Eur J Hum Genet* 2014, **22**(4):471-479 [doi.org/10.1038/ejhg.2013.189.](https://doi.org/10.1038/ejhg.2013.189.)

36. De Rocker N, Vergult S, Koolen D, Jacobs E, Hoischen A, Zeesman S, Bang B, Béna F, Bockaert N, Bongers EM *et al*: **Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity**. *Genet Med* 2015, **17**(6):460-466 [doi.org/10.1038/gim.2014.124.](https://doi.org/10.1038/gim.2014.124.)

37. Béna F, Bruno DL, Eriksson M, van Ravenswaaij-Arts C, Stark Z, Dijkhuizen T, Gerkes E, Gimelli S, Ganesamoorthy D, Thuresson AC *et al*: **Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature**. *American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics* 2013, **162b**(4):388-403 [doi.org/10.1002/ajmg.b.32148.](https://doi.org/10.1002/ajmg.b.32148.)

38. Lowther C, Speevak M, Armour CM, Goh ES, Graham GE, Li C, Zeesman S, Nowaczyk MJM, Schultz L-A, Morra A *et al*: **Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression**. *Genetics in Medicine* 2017, **19**(1):53-61 [doi.org/10.1038/gim.2016.54.](https://doi.org/10.1038/gim.2016.54.)

39. Al Shehhi M, Forman EB, Fitzgerald JE, McInerney V, Krawczyk J, Shen S, Betts DR, Ardle LM, Gorman KM, King MD *et al*: **NRXN1 deletion syndrome; phenotypic and penetrance data from 34 families**. *European journal of medical genetics* 2019, **62**(3):204-209 [doi.org/doi.org/10.1016/j.ejmg.2018.07.015.](https://doi.org/https://doi.org/10.1016/j.ejmg.2018.07.015.)

40. Kaminsky EB, Kaul V, Paschall J, Church DM, Bunke B, Kunig D, Moreno-De-Luca D, Moreno-De-Luca A, Mulle JG, Warren ST *et al*: **An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities**. *Genet Med* 2011, **13**(9):777-784 [doi.org/10.1097/GIM.0b013e31822c79f9.](https://doi.org/10.1097/GIM.0b013e31822c79f9.)

41. Piccione M, Piro E, Serraino F, Cavani S, Ciccone R, Malacarne M, Pierluigi M, Vitaloni M, Zuffardi O, Corsello G: **Interstitial deletion of chromosome 2p15-16.1: Report of two patients and critical review of current genotype–phenotype correlation**. *European journal of medical genetics* 2012, **55**(4):238-244 [doi.org/doi.org/10.1016/j.ejmg.2012.01.014.](https://doi.org/https://doi.org/10.1016/j.ejmg.2012.01.014.)

42. Hancarova M, Simandlova M, Drabova J, Mannik K, Kurg A, Sedlacek Z: **A patient with de novo 0.45 Mb deletion of 2p16.1: The role of BCL11A, PAPOLG, REL, and FLJ16341 in the 2p15-p16.1 microdeletion syndrome**. *American Journal of Medical Genetics Part A* 2013, **161**(4):865-870 [doi.org/doi.org/10.1002/ajmg.a.35783.](https://doi.org/https://doi.org/10.1002/ajmg.a.35783.)

43. Mimouni-Bloch A, Yeshaya J, Kahana S, Maya I, Basel-Vanagaite L: **A de-novo interstitial microduplication involving 2p16.1-p15 and mirroring 2p16.1-p15 microdeletion syndrome: Clinical and molecular analysis**. *European Journal of Paediatric Neurology* 2015, **19**(6):711-715 [doi.org/doi.org/10.1016/j.ejpn.2015.07.013.](https://doi.org/https://doi.org/10.1016/j.ejpn.2015.07.013.)

44. Prontera P, Bernardini L, Stangoni G, Capalbo A, Rogaia D, Romani R, Ardisia C, Dallapiccola B, Donti E: **Deletion 2p15-16.1 syndrome: case report and review**. *American journal of medical genetics Part A* 2011, **155a**(10):2473-2478 [doi.org/10.1002/ajmg.a.33875.](https://doi.org/10.1002/ajmg.a.33875.)

45. Wohlleber E, Kirchhoff M, Zink AM, Kreiss-Nachtsheim M, Küchler A, Jepsen B, Kjaergaard S, Engels H: **Clinical and molecular characterization of two patients with overlapping de novo microdeletions in 2p14-p15 and mild mental retardation**. *European journal of medical genetics* 2011, **54**(1):67-72 [doi.org/10.1016/j.ejmg.2010.09.012.](https://doi.org/10.1016/j.ejmg.2010.09.012.)

46. Hancarova M, Vejvalkova S, Trkova M, Drabova J, Dleskova A, Vlckova M, Sedlacek Z: **Identification of a patient with intellectual disability and de novo 3.7Mb deletion supports the existence of a novel microdeletion syndrome in 2p14–p15**. *Gene* 2013, **516**(1):158-161 [doi.org/doi.org/10.1016/j.gene.2012.12.027.](https://doi.org/https://doi.org/10.1016/j.gene.2012.12.027.)

47. Mathieu ML, Demily C, Chantot-Bastaraud S, Afenjar A, Mignot C, Andrieux J, Gerard M, Catala-Mora J, Jouk PS, Labalme A *et al*: **Clinical and molecular cytogenetic characterization of four unrelated patients carrying 2p14 microdeletions**. *American journal of medical genetics Part A* 2017, **173**(8):2268-2274 [doi.org/10.1002/ajmg.a.38307.](https://doi.org/10.1002/ajmg.a.38307.)

48. Pavone P, Falsaperla R, Rizzo R, Praticò AD, Ruggieri M: **Chromosome 2p15-p16.1 microduplication in a boy with congenital anomalies: Is it a distinctive syndrome?** *European journal of medical genetics* 2019, **62**(1):47-54 [doi.org/doi.org/10.1016/j.ejmg.2018.05.001.](https://doi.org/https://doi.org/10.1016/j.ejmg.2018.05.001.)

49. Barber JC, Thomas NS, Collinson MN, Dennis NR, Liehr T, Weise A, Belitz B, Pfeiffer L, Kirchhoff M, Krag-Olsen B *et al*: **Segmental haplosufficiency: transmitted deletions of 2p12 include a pancreatic regeneration gene cluster and have no apparent phenotypic consequences**. *Eur J Hum Genet* 2005, **13**(3):283-291 [doi.org/10.1038/sj.ejhg.5201267.](https://doi.org/10.1038/sj.ejhg.5201267.)

50. Tzschach A, Graul-Neumann LM, Konrat K, Richter R, Ebert G, Ullmann R, Neitzel H: **Interstitial deletion 2p11.2-p12: report of a patient with mental retardation and review of the literature**. *American journal of medical genetics Part A* 2009, **149a**(2):242-245 [doi.org/10.1002/ajmg.a.32637.](https://doi.org/10.1002/ajmg.a.32637.)

51. Writzl K, Lovrecić L, Peterlin B: **Interstitial deletion 2p11.2-p12: further delineation**. *American journal of medical genetics Part A* 2009, **149a**(10):2324-2326 [doi.org/10.1002/ajmg.a.33064.](https://doi.org/10.1002/ajmg.a.33064.)

52. Riley KN, Catalano LM, Bernat JA, Adams SD, Martin DM, Lalani SR, Patel A, Burnside RD, Innis JW, Rudd MK: **Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes**. *American journal of medical genetics Part A* 2015, **167a**(11):2664-2673 [doi.org/10.1002/ajmg.a.37269.](https://doi.org/10.1002/ajmg.a.37269.)

53. Wolfe K, McQuillin A, Alesi V, Boudry Labis E, Cutajar P, Dallapiccola B, Dentici ML, Dieux-Coeslier A, Duban-Bedu B, Duelund Hjortshøj T *et al*: **Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications**. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* 2018, **177**(4):397-405 [doi.org/doi.org/10.1002/ajmg.b.32627.](https://doi.org/https://doi.org/10.1002/ajmg.b.32627.)

54. Wilson M, Mowat D, Dastot-Le Moal F, Cacheux V, Kääriäinen H, Cass D, Donnai D, Clayton-Smith J, Townshend S, Curry C *et al*: **Further delineation of the phenotype associated with heterozygous mutations in ZFHX1B**. *American journal of medical genetics Part A* 2003, **119a**(3):257-265 [doi.org/10.1002/ajmg.a.20053.](https://doi.org/10.1002/ajmg.a.20053.)

55. Zweier C, Temple IK, Beemer F, Zackai E, Lerman-Sagie T, Weschke B, Anderson CE, Rauch A: **Characterisation of deletions of the ZFHX1B region and genotype-phenotype analysis in Mowat-Wilson syndrome**. *Journal of medical genetics* 2003, **40**(8):601-605 [doi.org/10.1136/jmg.40.8.601.](https://doi.org/10.1136/jmg.40.8.601.)

56. Ishihara N, Yamada K, Yamada Y, Miura K, Kato J, Kuwabara N, Hara Y, Kobayashi Y, Hoshino K, Nomura Y *et al*: **Clinical and molecular analysis of Mowat-Wilson syndrome associated with ZFHX1B mutations and deletions at 2q22-q24.1**. *Journal of medical genetics* 2004, **41**(5):387-393 [doi.org/10.1136/jmg.2003.016154.](https://doi.org/10.1136/jmg.2003.016154.)

57. Ivanovski I, Djuric O, Caraffi SG, Santodirocco D, Pollazzon M, Rosato S, Cordelli DM, Abdalla E, Accorsi P, Adam MP *et al*: **Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care**. *Genet Med* 2018, **20**(9):965-975 [doi.org/10.1038/gim.2017.221.](https://doi.org/10.1038/gim.2017.221.)

58. Jaillard S, Dubourg C, Gérard-Blanluet M, Delahaye A, Pasquier L, Dupont C, Henry C, Tabet AC, Lucas J, Aboura A *et al*: **2q23.1 microdeletion identified by array comparative genomic hybridisation: an emerging phenotype with Angelman-like features?** *Journal of medical genetics* 2009, **46**(12):847-855 [doi.org/10.1136/jmg.2008.058156.](https://doi.org/10.1136/jmg.2008.058156.)

59. van Bon BWM, Koolen DA, Brueton L, McMullan D, Lichtenbelt KD, Adès LC, Peters G, Gibson K, Moloney S, Novara F *et al*: **The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype**. *Eur J Hum Genet* 2010, **18**(2):163-170 [doi.org/10.1038/ejhg.2009.152.](https://doi.org/10.1038/ejhg.2009.152.)

60. Williams SR, Mullegama SV, Rosenfeld JA, Dagli AI, Hatchwell E, Allen WP, Williams CA, Elsea SH: **Haploinsufficiency of MBD5 associated with a syndrome involving microcephaly, intellectual disabilities, severe speech impairment, and seizures**. *Eur J Hum Genet* 2010, **18**(4):436-441 [doi.org/10.1038/ejhg.2009.199.](https://doi.org/10.1038/ejhg.2009.199.)

61. Talkowski ME, Mullegama SV, Rosenfeld JA, van Bon BWM, Shen Y, Repnikova EA, Gastier-Foster J, Thrush DL, Kathiresan S, Ruderfer DM *et al*: **Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder**. *American journal of human genetics* 2011, **89**(4):551-563 [doi.org/10.1016/j.ajhg.2011.09.011.](https://doi.org/10.1016/j.ajhg.2011.09.011.)

62. Cho TJ, Kim OH, Choi IH, Nishimura G, Superti-Furga A, Kim KS, Lee YJ, Park WY: **A dominant mesomelic dysplasia associated with a 1.0-Mb microduplication of HOXD gene cluster at 2q31.1**. *Journal of medical genetics* 2010, **47**(9):638-639 [doi.org/10.1136/jmg.2009.074690.](https://doi.org/10.1136/jmg.2009.074690.)

63. Kantaputra PN, Klopocki E, Hennig BP, Praphanphoj V, Le Caignec C, Isidor B, Kwee ML, Shears DJ, Mundlos S: **Mesomelic dysplasia Kantaputra type is associated with duplications of the HOXD locus on chromosome 2q**. *Eur J Hum Genet* 2010, **18**(12):1310-1314 [doi.org/10.1038/ejhg.2010.116.](https://doi.org/10.1038/ejhg.2010.116.)

64. Mitter D, Chiaie BD, Lüdecke H-J, Gillessen-Kaesbach G, Bohring A, Kohlhase J, Caliebe A, Siebert R, Roepke A, Ramos-Arroyo MA *et al*: **Genotype–phenotype correlation in eight new patients with a deletion encompassing 2q31.1**. *American Journal of Medical Genetics Part A* 2010, **152A**(5):1213-1224 [doi.org/doi.org/10.1002/ajmg.a.33344.](https://doi.org/https://doi.org/10.1002/ajmg.a.33344.)

65. Theisen A, Rosenfeld JA, Shane K, McBride KL, Atkin JF, Gaba C, Hoo J, Kurczynski TW, Schnur RE, Coffey LB *et al*: **Refinement of the Region for Split Hand/Foot Malformation 5 on 2q31.1**. *Molecular Syndromology* 2010, **1**(5):262-271 [doi.org/10.1159/000328405.](https://doi.org/10.1159/000328405.)

66. Dimitrov B, Balikova I, de Ravel T, Van Esch H, De Smedt M, Baten E, Vermeesch JR, Bradinova I, Simeonov E, Devriendt K *et al*: **2q31.1 microdeletion syndrome: redefining the associated clinical phenotype**. *Journal of medical genetics* 2011, **48**(2):98 [doi.org/10.1136/jmg.2010.079491.](https://doi.org/10.1136/jmg.2010.079491.)

67. Ghoumid J, Andrieux J, Sablonnière B, Odent S, Philippe N, Zanlonghi X, Saugier-Veber P, Bardyn T, Manouvrier-Hanu S, Holder-Espinasse M: **Duplication at chromosome 2q31.1-q31.2 in a family presenting syndactyly and nystagmus**. *European Journal of Human Genetics* 2011, **19**(11):1198-1201 [doi.org/10.1038/ejhg.2011.95.](https://doi.org/10.1038/ejhg.2011.95.)

68. Gurrieri F, Everman DB: **Clinical, genetic, and molecular aspects of split-hand/foot malformation: An update**. *American Journal of Medical Genetics Part A* 2013, **161**(11):2860-2872 [doi.org/doi.org/10.1002/ajmg.a.36239.](https://doi.org/https://doi.org/10.1002/ajmg.a.36239.)

69. Sowińska-Seidler A, Socha M, Jamsheer A: **Split-hand/foot malformation - molecular cause and implications in genetic counseling**. *Journal of Applied Genetics* 2014, **55**(1):105-115 [doi.org/10.1007/s13353-013-0178-5.](https://doi.org/10.1007/s13353-013-0178-5.)

70. Prontera P, Bernardini L, Stangoni G, Capalbo A, Rogaia D, Ardisia C, Novelli A, Dallapiccola B, Donti E: **2q31.2q32.3 deletion syndrome: report of an adult patient**. *American journal of medical genetics Part A* 2009, **149a**(4):706-712 [doi.org/10.1002/ajmg.a.32688.](https://doi.org/10.1002/ajmg.a.32688.)

71. Cocchella A, Malacarne M, Forzano F, Marciano C, Pierluigi M, Perroni L, Faravelli F, Di Maria E: **The refinement of the critical region for the 2q31.2q32.3 deletion syndrome indicates candidate genes for mental retardation and speech impairment**. *American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics* 2010, **153b**(7):1342-1346 [doi.org/10.1002/ajmg.b.31107.](https://doi.org/10.1002/ajmg.b.31107.)

72. Manolakos E, Vetro A, Kefalas K, Rapti SM, Louizou E, Garas A, Kitsos G, Vasileiadis L, Tsoplou P, Eleftheriades M *et al*: **The use of array-CGH in a cohort of Greek children with developmental delay**. *Molecular cytogenetics* 2010, **3**:22 [doi.org/10.1186/1755-8166-3-22.](https://doi.org/10.1186/1755-8166-3-22.)

73. Ferreira SI, Matoso E, Venâncio M, Saraiva J, Melo JB, Carreira IM: **Critical region in 2q31.2q32.3 deletion syndrome: Report of two phenotypically distinct patients, one with an additional deletion in Alagille syndrome region**. *Molecular cytogenetics* 2012, **5**(1):25-25 [doi.org/10.1186/1755-8166-5-25.](https://doi.org/10.1186/1755-8166-5-25.)

74. Rosenfeld JA, Ballif BC, Lucas A, Spence EJ, Powell C, Aylsworth AS, Torchia BA, Shaffer LG: **Small Deletions of SATB2 Cause Some of the Clinical Features of the 2q33.1 Microdeletion Syndrome**. *PLOS ONE* 2009, **4**(8):e6568 [doi.org/10.1371/journal.pone.0006568.](https://doi.org/10.1371/journal.pone.0006568.)

75. Balasubramanian M, Smith K, Basel-Vanagaite L, Feingold MF, Brock P, Gowans GC, Vasudevan PC, Cresswell L, Taylor EJ, Harris CJ *et al*: **Case series: 2q33.1 microdeletion syndrome—further delineation of the phenotype**. *Journal of medical genetics* 2011, **48**(5):290 [doi.org/10.1136/jmg.2010.084491.](https://doi.org/10.1136/jmg.2010.084491.)

76. Zarate YA, Fish JL: **SATB2-associated syndrome: Mechanisms, phenotype, and practical recommendations**. *American Journal of Medical Genetics Part A* 2017, **173**(2):327-337 [doi.org/doi.org/10.1002/ajmg.a.38022.](https://doi.org/https://doi.org/10.1002/ajmg.a.38022.)

77. Klopocki E, Lohan S, Brancati F, Koll R, Brehm A, Seemann P, Dathe K, Stricker S, Hecht J, Bosse K *et al*: **Copy-number variations involving the IHH locus are associated with syndactyly and craniosynostosis**. *American journal of human genetics* 2011, **88**(1):70-75 [doi.org/10.1016/j.ajhg.2010.11.006.](https://doi.org/10.1016/j.ajhg.2010.11.006.)

78. Yuksel-Apak M, Bögershausen N, Pawlik B, Li Y, Apak S, Uyguner O, Milz E, Nürnberg G, Karaman B, Gülgören A *et al*: **A large duplication involving the IHH locus mimics acrocallosal syndrome**. *European Journal of Human Genetics* 2012, **20**(6):639-644 [doi.org/10.1038/ejhg.2011.250.](https://doi.org/10.1038/ejhg.2011.250.)

79. Spielmann M, Klopocki E: **CNVs of noncoding cis-regulatory elements in human disease**. *Current Opinion in Genetics & Development* 2013, **23**(3):249-256 [doi.org/doi.org/10.1016/j.gde.2013.02.013.](https://doi.org/https://doi.org/10.1016/j.gde.2013.02.013.)

80. Barroso E, Berges-Soria J, Benito-Sanz S, Rivera-Pedroza CI, Ballesta-Martínez MJ, López-González V, Guillen-Navarro E, Heath KE: **Identification of the fourth duplication of upstream IHH regulatory elements, in a family with craniosynostosis Philadelphia type, helps to define the phenotypic characterization of these regulatory elements**. *American journal of medical genetics Part A* 2015, **167a**(4):902-906 [doi.org/10.1002/ajmg.a.36811.](https://doi.org/10.1002/ajmg.a.36811.)

81. Williams SR, Aldred MA, Der Kaloustian VM, Halal F, Gowans G, McLeod DR, Zondag S, Toriello HV, Magenis RE, Elsea SH: **Haploinsufficiency of HDAC4 causes brachydactyly mental retardation syndrome, with brachydactyly type E, developmental delays, and behavioral problems**. *American journal of human genetics* 2010, **87**(2):219-228 [doi.org/10.1016/j.ajhg.2010.07.011.](https://doi.org/10.1016/j.ajhg.2010.07.011.)

82. Villavicencio-Lorini P, Klopocki E, Trimborn M, Koll R, Mundlos S, Horn D: **Phenotypic variant of Brachydactyly-mental retardation syndrome in a family with an inherited interstitial 2q37.3 microdeletion including HDAC4**. *Eur J Hum Genet* 2013, **21**(7):743-748 [doi.org/10.1038/ejhg.2012.240.](https://doi.org/10.1038/ejhg.2012.240.)

83. Wheeler PG, Huang D, Dai Z: **Haploinsufficiency of HDAC4 does not cause intellectual disability in all affected individuals**. *American journal of medical genetics Part A* 2014, **164a**(7):1826-1829 [doi.org/10.1002/ajmg.a.36542.](https://doi.org/10.1002/ajmg.a.36542.)

84. Jean-Marçais N, Decamp M, Gérard M, Ribault V, Andrieux J, Kottler ML, Plessis G: **The first familial case of inherited 2q37.3 interstitial deletion with isolated skeletal abnormalities including brachydactyly type E and short stature**. *American journal of medical genetics Part A* 2015, **167a**(1):185-189 [doi.org/10.1002/ajmg.a.36428.](https://doi.org/10.1002/ajmg.a.36428.)

85. Le TN, Williams SR, Alaimo JT, Elsea SH: **Genotype and phenotype correlation in 103 individuals with 2q37 deletion syndrome reveals incomplete penetrance and supports HDAC4 as the primary genetic contributor**. *American journal of medical genetics Part A* 2019, **179**(5):782-791 [doi.org/10.1002/ajmg.a.61089.](https://doi.org/10.1002/ajmg.a.61089.)

86. Gunnarsson C, Foyn Bruun C: **Molecular characterization and clinical features of a patient with an interstitial deletion of 3p25.3-p26.1**. *American journal of medical genetics Part A* 2010, **152a**(12):3110-3114 [doi.org/10.1002/ajmg.a.33353.](https://doi.org/10.1002/ajmg.a.33353.)

87. Bownass L, Lunt P, Williams C, Woodbridge T, Ellis M, Scurr I: **A report of a new case of an interstitial deletion at 3p25.3 and expansion of the clinical phenotype**. *Clinical dysmorphology* 2014, **23**(2):63-66 [doi.org/10.1097/mcd.0000000000000030.](https://doi.org/10.1097/mcd.0000000000000030.)

88. Dikow N, Maas B, Karch S, Granzow M, Janssen JW, Jauch A, Hinderhofer K, Sutter C, Schubert-Bast S, Anderlid BM *et al*: **3p25.3 microdeletion of GABA transporters SLC6A1 and SLC6A11 results in intellectual disability, epilepsy and stereotypic behavior**. *American journal of medical genetics Part A* 2014, **164a**(12):3061-3068 [doi.org/10.1002/ajmg.a.36761.](https://doi.org/10.1002/ajmg.a.36761.)

89. Kuechler A, Zink AM, Wieland T, Lüdecke H-J, Cremer K, Salviati L, Magini P, Najafi K, Zweier C, Czeschik JC *et al*: **Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome**. *European Journal of Human Genetics* 2015, **23**(6):753-760 [doi.org/10.1038/ejhg.2014.165.](https://doi.org/10.1038/ejhg.2014.165.)

90. Haldeman-Englert CR, Gai X, Perin JC, Ciano M, Halbach SS, Geiger EA, McDonald-McGinn DM, Hakonarson H, Zackai EH, Shaikh TH: **A 3.1-Mb microdeletion of 3p21.31 associated with cortical blindness, cleft lip, CNS abnormalities, and developmental delay**. *European journal of medical genetics* 2009, **52**(4):265-268 [doi.org/10.1016/j.ejmg.2008.11.005.](https://doi.org/10.1016/j.ejmg.2008.11.005.)

91. Eto K, Sakai N, Shimada S, Shioda M, Ishigaki K, Hamada Y, Shinpo M, Azuma J, Tominaga K, Shimojima K *et al*: **Microdeletions of 3p21.31 characterized by developmental delay, distinctive features, elevated serum creatine kinase levels, and white matter involvement**. *American journal of medical genetics Part A* 2013, **161a**(12):3049-3056 [doi.org/10.1002/ajmg.a.36156.](https://doi.org/10.1002/ajmg.a.36156.)

92. Lovrecic L, Bertok S, Žerjav Tanšek M: **A New Case of an Extremely Rare 3p21.31 Interstitial Deletion**. *Molecular syndromology* 2016, **7**(2):93-98 [doi.org/10.1159/000445227.](https://doi.org/10.1159/000445227.)

93. Molin AM, Andrieux J, Koolen DA, Malan V, Carella M, Colleaux L, Cormier-Daire V, David A, de Leeuw N, Delobel B *et al*: **A novel microdeletion syndrome at 3q13.31 characterised by developmental delay, postnatal overgrowth, hypoplastic male genitals, and characteristic facial features**. *Journal of medical genetics* 2012, **49**(2):104-109 [doi.org/10.1136/jmedgenet-2011-100534.](https://doi.org/10.1136/jmedgenet-2011-100534.)

94. Vuillaume M-L, Delrue M-A, Naudion S, Toutain J, Fergelot P, Arveiler B, Lacombe D, Rooryck C: **Expanding the clinical phenotype at the 3q13.31 locus with a new case of microdeletion and first characterization of the reciprocal duplication**. *Molecular Genetics and Metabolism* 2013, **110**(1):90-97 [doi.org/doi.org/10.1016/j.ymgme.2013.07.013.](https://doi.org/https://doi.org/10.1016/j.ymgme.2013.07.013.)

95. Hervé B, Fauvert D, Dard R, Roume J, Cognard S, Goidin D, Lozach F, Molina-Gomes D, Vialard F: **The emerging microduplication 3q13.31: Expanding the genotype-phenotype correlations of the reciprocal microdeletion 3q13.31 syndrome**. *European journal of medical genetics* 2016, **59**(9):463-469 [doi.org/doi.org/10.1016/j.ejmg.2016.08.010.](https://doi.org/https://doi.org/10.1016/j.ejmg.2016.08.010.)

96. Lim BC, Park WY, Seo E-J, Kim KJ, Hwang YS, Chae JH: **De Novo Interstitial Deletion of 3q22.3-q25.2 Encompassing FOXL2, ATR, ZIC1, and ZIC4 in a Patient With Blepharophimosis/Ptosis/Epicanthus Inversus Syndrome, Dandy-Walker Malformation, and Global Developmental Delay**. *Journal of Child Neurology* 2011, **26**(5):615-618 [doi.org/10.1177/0883073810384996.](https://doi.org/10.1177/0883073810384996.)

97. Moortgat S, Verellen-Dumoulin C, Maystadt I, Parmentier B, Grisart B, Hennecker JL, Destree A: **Developmental delay and facial dysmorphism in a child with an 8.9 Mb de novo interstitial deletion of 3q25.1-q25.32: Genotype-phenotype correlations of chromosome 3q25 deletion syndrome**. *European journal of medical genetics* 2011, **54**(2):177-180 [doi.org/10.1016/j.ejmg.2010.11.011.](https://doi.org/10.1016/j.ejmg.2010.11.011.)

98. Ferraris A, Bernardini L, Sabolic Avramovska V, Zanni G, Loddo S, Sukarova-Angelovska E, Parisi V, Capalbo A, Tumini S, Travaglini L *et al*: **Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions**. *Orphanet J Rare Dis* 2013, **8**(1):75 [doi.org/10.1186/1750-1172-8-75.](https://doi.org/10.1186/1750-1172-8-75.)

99. Bertini V, Orsini A, Mazza R, Mandava V, Saggese G, Azzara’ A, Bonuccelli A, Valetto A: **A 6.5 mb deletion at 3q24q25.2 narrows Wisconsin syndrome critical region to a 750 kb interval: A potential role for MBNLI**. *American Journal of Medical Genetics Part A* 2017, **173**(1):280-284 [doi.org/doi.org/10.1002/ajmg.a.38002.](https://doi.org/https://doi.org/10.1002/ajmg.a.38002.)

100. Ramineni A, Coman D: **De Novo 3q22.3q24 Microdeletion in a Patient With Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome, Dandy-Walker Malformation, and Wisconsin Syndrome**. *Child neurology open* 2016, **3**:2329048x16666362 [doi.org/10.1177/2329048x16666362.](https://doi.org/10.1177/2329048x16666362.)

101. Thevenon J, Callier P, Poquet H, Bache I, Menten B, Malan V, Cavaliere ML, Girod JP, Thauvin-Robinet C, El Chehadeh S *et al*: **3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder**. *Journal of medical genetics* 2014, **51**(1):21-27 [doi.org/10.1136/jmedgenet-2013-101939.](https://doi.org/10.1136/jmedgenet-2013-101939.)

102. Ponzi E, Asaro A, Orteschi D, Genuardi M, Zollino M, Gurrieri F: **Variable expressivity of a familial 1.9 Mb microdeletion in 3q28 leading to haploinsufficiency of TP63: Refinement of the critical region for a new microdeletion phenotype**. *European journal of medical genetics* 2015, **58**(8):400-405 [doi.org/doi.org/10.1016/j.ejmg.2015.06.001.](https://doi.org/https://doi.org/10.1016/j.ejmg.2015.06.001.)

103. Maas NMC, Van Buggenhout G, Hannes F, Thienpont B, Sanlaville D, Kok K, Midro A, Andrieux J, Anderlid BM, Schoumans J *et al*: **Genotype–phenotype correlation in 21 patients with Wolf–Hirschhorn syndrome using high resolution array comparative genome hybridisation (CGH)**. *Journal of medical genetics* 2008, **45**(2):71 [doi.org/10.1136/jmg.2007.052910.](https://doi.org/10.1136/jmg.2007.052910.)

104. Ho KS, South ST, Lortz A, Hensel CH, Sdano MR, Vanzo RJ, Martin MM, Peiffer A, Lambert CG, Calhoun A *et al*: **Chromosomal microarray testing identifies a 4p terminal region associated with seizures in Wolf-Hirschhorn syndrome**. *Journal of medical genetics* 2016, **53**(4):256-263 [doi.org/10.1136/jmedgenet-2015-103626.](https://doi.org/10.1136/jmedgenet-2015-103626.)

105. Corrêa T, Mergener R, Leite JC, Galera MF, Moreira LM, Vargas JE, Riegel M: **Cytogenomic Integrative Network Analysis of the Critical Region Associated with Wolf-Hirschhorn Syndrome**. *BioMed Research International* 2018, **2018** [doi.org/10.1155/2018/5436187.](https://doi.org/10.1155/2018/5436187.)

106. Harada N, Nagai T, Shimokawa O, Niikawa N, Matsumoto N: **A 4q21-q22 deletion in a girl with severe growth retardation**. *Clin Genet* 2002, **61**(3):226-228 [doi.org/10.1034/j.1399-0004.2002.610311.x.](https://doi.org/10.1034/j.1399-0004.2002.610311.x.)

107. Bonnet C, Andrieux J, Béri-Dexheimer M, Leheup B, Boute O, Manouvrier S, Delobel B, Copin H, Receveur A, Mathieu M *et al*: **Microdeletion at chromosome 4q21 defines a new emerging syndrome with marked growth restriction, mental retardation and absent or severely delayed speech**. *Journal of medical genetics* 2010, **47**(6):377-384 [doi.org/10.1136/jmg.2009.071902.](https://doi.org/10.1136/jmg.2009.071902.)

108. Dukes-Rimsky L, Guzauskas GF, Holden KR, Griggs R, Ladd S, Montoya MdC, DuPont BR, Srivastava AK: **Microdeletion at 4q21.3 is associated with intellectual disability, dysmorphic facies, hypotonia, and short stature**. *American journal of medical genetics Part A* 2011, **155A**(9):2146-2153 [doi.org/10.1002/ajmg.a.34137.](https://doi.org/10.1002/ajmg.a.34137.)

109. Hu X, Chen X, Wu B, Soler IM, Chen S, Shen Y: **Further defining the critical genes for the 4q21 microdeletion disorder**. *American Journal of Medical Genetics Part A* 2017, **173**(1):120-125 [doi.org/doi.org/10.1002/ajmg.a.37965.](https://doi.org/https://doi.org/10.1002/ajmg.a.37965.)

110. Yan J, Zhang F, Brundage E, Scheuerle A, Lanpher B, Erickson RP, Powis Z, Robinson HB, Trapane PL, Stachiw-Hietpas D *et al*: **Genomic duplication resulting in increased copy number of genes encoding the sister chromatid cohesion complex conveys clinical consequences distinct from Cornelia de Lange**. *Journal of medical genetics* 2009, **46**(9):626-634 [doi.org/10.1136/jmg.2008.062471.](https://doi.org/10.1136/jmg.2008.062471.)

111. Romero MCC, Hoyo RG, Calvente M, Cano MB, Castillo LG, Suela J: **Neonatal detection of 5p13.2 duplication and delineation of the phenotype**. *American Journal of Medical Genetics Part A* 2012, **158A**(4):877-881 [doi.org/doi.org/10.1002/ajmg.a.35237.](https://doi.org/https://doi.org/10.1002/ajmg.a.35237.)

112. Lucarelli E, Pasca MG, Fanizza I, Trabacca A: **Electroclinical characteristics and neuropsychological profile of a female child with chromosome 5p13.2 duplication syndrome**. *Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology* 2017, **38**(5):915-917 [doi.org/10.1007/s10072-017-2825-9.](https://doi.org/10.1007/s10072-017-2825-9.)

113. Jaillard S, Andrieux J, Plessis G, Krepischi AC, Lucas J, David V, Le Brun M, Bertola DR, David A, Belaud-Rotureau MA *et al*: **5q12.1 deletion: delineation of a phenotype including mental retardation and ocular defects**. *American journal of medical genetics Part A* 2011, **155a**(4):725-731 [doi.org/10.1002/ajmg.a.33758.](https://doi.org/10.1002/ajmg.a.33758.)

114. Lindstrand A, Grigelioniene G, Nilsson D, Pettersson M, Hofmeister W, Anderlid BM, Kant SG, Ruivenkamp CA, Gustavsson P, Valta H *et al*: **Different mutations in PDE4D associated with developmental disorders with mirror phenotypes**. *Journal of medical genetics* 2014, **51**(1):45-54 [doi.org/10.1136/jmedgenet-2013-101937.](https://doi.org/10.1136/jmedgenet-2013-101937.)

115. Snijders Blok C, Corsten-Janssen N, FitzPatrick DR, Romano C, Fichera M, Vitello GA, Willemsen MH, Schoots J, Pfundt R, van Ravenswaaij-Arts CM *et al*: **Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes**. *American journal of medical genetics Part A* 2014, **164a**(11):2843-2848 [doi.org/10.1002/ajmg.a.36680.](https://doi.org/10.1002/ajmg.a.36680.)

116. Fontana P, Tortora C, Petillo R, Falco M, Miniero M, De Brasi D, Pisanti MA: **A novel 5q11.2 microdeletion in a child with mild developmental delay and dysmorphic features**. *American journal of medical genetics Part A* 2016, **170**(9):2445-2448 [doi.org/10.1002/ajmg.a.37824.](https://doi.org/10.1002/ajmg.a.37824.)

117. Arora V, Aggarwal S, Bijarnia S, Lall M, Joshi A, Dua-Puri R, Arora U, Verma I: **Extending the Phenotype and Identification of a Novel Candidate Gene for Immunodeficiency in 5q11 Microdeletion Syndrome**. *Molecular syndromology* 2019, **9**(6):312-318 [doi.org/10.1159/000494995.](https://doi.org/10.1159/000494995.)

118. Cellamare A, Coccaro N, Nuzzi MC, Casieri P, Tampoia M, Maggiolini FA, Gentile M, Ficarella R, Ponzi E, Conserva MR *et al*: **Cytogenetic and Array-CGH Characterization of a Simple Case of Reciprocal t(3;10) Translocation Reveals a Hidden Deletion at 5q12**. *Genes* 2021, **12**(6) [doi.org/10.3390/genes12060877.](https://doi.org/10.3390/genes12060877.)

119. Cardoso C, Boys A, Parrini E, Mignon-Ravix C, McMahon JM, Khantane S, Bertini E, Pallesi E, Missirian C, Zuffardi O *et al*: **Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion**. *Neurology* 2009, **72**(9):784-792 [doi.org/10.1212/01.wnl.0000336339.08878.2d.](https://doi.org/10.1212/01.wnl.0000336339.08878.2d.)

120. Engels H, Wohlleber E, Zink A, Hoyer J, Ludwig KU, Brockschmidt FF, Wieczorek D, Moog U, Hellmann-Mersch B, Weber RG *et al*: **A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients**. *Eur J Hum Genet* 2009, **17**(12):1592-1599 [doi.org/10.1038/ejhg.2009.90.](https://doi.org/10.1038/ejhg.2009.90.)

121. Sobreira N, Walsh MF, Batista D, Wang T: **Interstitial deletion 5q14.3-q21 associated with iris coloboma, hearing loss, dental anomaly, moderate intellectual disability, and attention deficit and hyperactivity disorder**. *American journal of medical genetics Part A* 2009, **149a**(11):2581-2583 [doi.org/10.1002/ajmg.a.33079.](https://doi.org/10.1002/ajmg.a.33079.)

122. Le Meur N, Holder-Espinasse M, Jaillard S, Goldenberg A, Joriot S, Amati-Bonneau P, Guichet A, Barth M, Charollais A, Journel H *et al*: **MEF2C haploinsufficiency caused by either microdeletion of the 5q14.3 region or mutation is responsible for severe mental retardation with stereotypic movements, epilepsy and/or cerebral malformations**. *Journal of medical genetics* 2010, **47**(1):22-29 [doi.org/10.1136/jmg.2009.069732.](https://doi.org/10.1136/jmg.2009.069732.)

123. Hotz A, Hellenbroich Y, Sperner J, Linder-Lucht M, Tacke U, Walter C, Caliebe A, Nagel I, Saunders DE, Wolff G *et al*: **Microdeletion 5q14.3 and anomalies of brain development**. *American journal of medical genetics Part A* 2013, **161a**(9):2124-2133 [doi.org/10.1002/ajmg.a.36020.](https://doi.org/10.1002/ajmg.a.36020.)

124. Brown KK, Alkuraya FS, Matos M, Robertson RL, Kimonis VE, Morton CC: **NR2F1 deletion in a patient with a de novo paracentric inversion, inv(5)(q15q33.2), and syndromic deafness**. *American journal of medical genetics Part A* 2009, **149a**(5):931-938 [doi.org/10.1002/ajmg.a.32764.](https://doi.org/10.1002/ajmg.a.32764.)

125. Al-Kateb H, Shimony JS, Vineyard M, Manwaring L, Kulkarni S, Shinawi M: **NR2F1 haploinsufficiency is associated with optic atrophy, dysmorphism and global developmental delay**. *American journal of medical genetics Part A* 2013, **161a**(2):377-381 [doi.org/10.1002/ajmg.a.35650.](https://doi.org/10.1002/ajmg.a.35650.)

126. Shimojima K, Isidor B, Le Caignec C, Kondo A, Sakata S, Ohno K, Yamamoto T: **A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination**. *American journal of medical genetics Part A* 2011, **155a**(4):732-736 [doi.org/10.1002/ajmg.a.33891.](https://doi.org/10.1002/ajmg.a.33891.)

127. Hosoki K, Ohta T, Natsume J, Imai S, Okumura A, Matsui T, Harada N, Bacino CA, Scaglia F, Jones JY *et al*: **Clinical phenotype and candidate genes for the 5q31.3 microdeletion syndrome**. *American journal of medical genetics Part A* 2012, **158a**(8):1891-1896 [doi.org/10.1002/ajmg.a.35439.](https://doi.org/10.1002/ajmg.a.35439.)

128. Brown N, Burgess T, Forbes R, McGillivray G, Kornberg A, Mandelstam S, Stark Z: **5q31.3 Microdeletion syndrome: clinical and molecular characterization of two further cases**. *American journal of medical genetics Part A* 2013, **161a**(10):2604-2608 [doi.org/10.1002/ajmg.a.36108.](https://doi.org/10.1002/ajmg.a.36108.)

129. Shimojima K, Okamoto N, Ohmura K, Nagase H, Yamamoto T: **Infantile spasms related to a 5q31.2-q31.3 microdeletion including PURA**. *Human Genome Variation* 2018, **5**(1):18007 [doi.org/10.1038/hgv.2018.7.](https://doi.org/10.1038/hgv.2018.7.)

130. Aldinger KA, Lehmann OJ, Hudgins L, Chizhikov VV, Bassuk AG, Ades LC, Krantz ID, Dobyns WB, Millen KJ: **FOXC1 is required for normal cerebellar development and is a major contributor to chromosome 6p25.3 Dandy-Walker malformation**. *Nature genetics* 2009, **41**(9):1037-1042 [doi.org/10.1038/ng.422.](https://doi.org/10.1038/ng.422.)

131. Delahaye A, Khung-Savatovsky S, Aboura A, Guimiot F, Drunat S, Alessandri J-L, Gérard M, Bitoun P, Boumendil J, Robin S *et al*: **Pre- and postnatal phenotype of 6p25 deletions involving the FOXC1 gene**. *American Journal of Medical Genetics Part A* 2012, **158A**(10):2430-2438 [doi.org/doi.org/10.1002/ajmg.a.35548.](https://doi.org/https://doi.org/10.1002/ajmg.a.35548.)

132. Vernon HJ, Bytyci Telegrafi A, Batista D, Owegi M, Leigh R: **6p25 microdeletion: white matter abnormalities in an adult patient**. *American journal of medical genetics Part A* 2013, **161a**(7):1686-1689 [doi.org/10.1002/ajmg.a.35937.](https://doi.org/10.1002/ajmg.a.35937.)

133. Linhares ND, Svartman M, Rodrigues TC, Rosenberg C, Valadares ER: **Subtelomeric 6p25 deletion/duplication: Report of a patient with new clinical findings and genotype–phenotype correlations**. *European journal of medical genetics* 2015, **58**(5):310-318 [doi.org/doi.org/10.1016/j.ejmg.2015.02.011.](https://doi.org/https://doi.org/10.1016/j.ejmg.2015.02.011.)

134. Kuipers BC, Vulto-van Silfhout AT, Marcelis C, Pfundt R, de Leeuw N, de Vries BB: **Two patients with intellectual disability, overlapping facial features, and overlapping deletions in 6p25.1p24.3**. *Clinical dysmorphology* 2013, **22**(1):18-21 [doi.org/10.1097/MCD.0b013e32835b6e39.](https://doi.org/10.1097/MCD.0b013e32835b6e39.)

135. Qi Z, Jeng LJB, Slavotinek A, Yu J: **Haploinsufficiency and triploinsensitivity of the same 6p25.1p24.3 region in a family**. *BMC Medical Genomics* 2015, **8**(1):38 [doi.org/10.1186/s12920-015-0113-1.](https://doi.org/10.1186/s12920-015-0113-1.)

136. Kent OA, Saha M, Coyaud E, Burston HE, Law N, Dadson K, Chen S, Laurent EM, St-Germain J, Sun RX *et al*: **Haploinsufficiency of RREB1 causes a Noonan-like RASopathy via epigenetic reprogramming of RAS-MAPK pathway genes**. *Nature communications* 2020, **11**(1):4673 [doi.org/10.1038/s41467-020-18483-9.](https://doi.org/10.1038/s41467-020-18483-9.)

137. Tassano E, Uccella S, Severino M, Giacomini T, Nardi F, Gimelli G, Tavella E, Ronchetto P, Malacarne M, Coviello D: **Expanding the phenotype associated with interstitial 6p25.1p24.3 microdeletion: a new case and review of the literature**. *Journal of Genetics* 2021, **100**(1):9 [doi.org/10.1007/s12041-021-01261-x.](https://doi.org/10.1007/s12041-021-01261-x.)

138. Wang JC, Dang L, Lomax B, Turner L, Shago M, Teebi AS, Klatt R, MacLeod PM, Yong SL, Nowaczyk MJ *et al*: **Molecular breakpoint mapping of 6q11-q14 interstitial deletions in seven patients**. *American journal of medical genetics Part A* 2009, **149a**(3):372-379 [doi.org/10.1002/ajmg.a.32675.](https://doi.org/10.1002/ajmg.a.32675.)

139. Engwerda A, Frentz B, den Ouden AL, Flapper BCT, Swertz MA, Gerkes EH, Plantinga M, Dijkhuizen T, van Ravenswaaij-Arts CMA: **The phenotypic spectrum of proximal 6q deletions based on a large cohort derived from social media and literature reports**. *European Journal of Human Genetics* 2018, **26**(10):1478-1489 [doi.org/10.1038/s41431-018-0172-9.](https://doi.org/10.1038/s41431-018-0172-9.)

140. Lowry RB, Chernos JE, Connelly MS, Wyse JPH: **Interstitial deletions at 6q14.1q15 associated with developmental delay and a marfanoid phenotype**. *Molecular syndromology* 2013, **4**(6):280-284 [doi.org/10.1159/000354038.](https://doi.org/10.1159/000354038.)

141. Dworschak GC, Draaken M, Hilger AC, Schramm C, Bartels E, Schmiedeke E, Grasshoff-Derr S, Märzheuser S, Holland-Cunz S, Lacher M *et al*: **Genome-wide mapping of copy number variations in patients with both anorectal malformations and central nervous system abnormalities**. *Birth defects research Part A, Clinical and molecular teratology* 2015, **103**(4):235-242 [doi.org/10.1002/bdra.23321.](https://doi.org/10.1002/bdra.23321.)

142. Van Esch H, Rosser EM, Janssens S, Van Ingelghem I, Loeys B, Menten B: **Developmental delay and connective tissue disorder in four patients sharing a common microdeletion at 6q13-14**. *Journal of medical genetics* 2010, **47**(10):717-720 [doi.org/10.1136/jmg.2010.077586.](https://doi.org/10.1136/jmg.2010.077586.)

143. Thienpont B, Zhang L, Postma AV, Breckpot J, Tranchevent L-C, Van Loo P, Møllgård K, Tommerup N, Bache I, Tümer Z *et al*: **Haploinsufficiency of TAB2 causes congenital heart defects in humans**. *American journal of human genetics* 2010, **86**(6):839-849 [doi.org/10.1016/j.ajhg.2010.04.011.](https://doi.org/10.1016/j.ajhg.2010.04.011.)

144. Nagamani SCS, Erez A, Eng C, Ou Z, Chinault C, Workman L, Coldwell J, Stankiewicz P, Patel A, Lupski JR *et al*: **Interstitial deletion of 6q25.2-q25.3: a novel microdeletion syndrome associated with microcephaly, developmental delay, dysmorphic features and hearing loss**. *Eur J Hum Genet* 2009, **17**(5):573-581 [doi.org/10.1038/ejhg.2008.220.](https://doi.org/10.1038/ejhg.2008.220.)

145. Santen GW, Clayton-Smith J: **The ARID1B phenotype: what we have learned so far**. *American journal of medical genetics Part C, Seminars in medical genetics* 2014, **166c**(3):276-289 [doi.org/10.1002/ajmg.c.31414.](https://doi.org/10.1002/ajmg.c.31414.)

146. Ronzoni L, Tagliaferri F, Tucci A, Baccarin M, Esposito S, Milani D: **Interstitial 6q25 microdeletion syndrome: ARID1B is the key gene**. *American Journal of Medical Genetics Part A* 2016, **170**(5):1257-1261 [doi.org/doi.org/10.1002/ajmg.a.37553.](https://doi.org/https://doi.org/10.1002/ajmg.a.37553.)

147. Zhong M-L, Song Y-M, Zou C-C: **6q25.1-q25.3 Microdeletion in a Chinese Girl**. *J Clin Res Pediatr Endocrinol* 2021, **13**(1):109-113 [doi.org/10.4274/jcrpe.galenos.2020.2020.0008.](https://doi.org/10.4274/jcrpe.galenos.2020.2020.0008.)

148. Caselli R, Ballarati L, Vignoli A, Peron A, Recalcati MP, Catusi I, Larizza L, Giardino D: **7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature**. *European journal of medical genetics* 2015, **58**(11):578-583 [doi.org/10.1016/j.ejmg.2015.08.003.](https://doi.org/10.1016/j.ejmg.2015.08.003.)

149. Shimojima K, Narai S, Togawa M, Doumoto T, Sangu N, Vanakker OM, de Paepe A, Edwards M, Whitehall J, Brescianini S *et al*: **7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly**. *European journal of medical genetics* 2016, **59**(10):502-506 [doi.org/10.1016/j.ejmg.2016.09.008.](https://doi.org/10.1016/j.ejmg.2016.09.008.)

150. Del Campo M, Antonell A, Magano LF, Muñoz FJ, Flores R, Bayés M, Pérez Jurado LA: **Hemizygosity at the NCF1 gene in patients with Williams-Beuren syndrome decreases their risk of hypertension**. *American journal of human genetics* 2006, **78**(4):533-542 [doi.org/10.1086/501073.](https://doi.org/10.1086/501073.)

151. Van der Aa N, Rooms L, Vandeweyer G, van den Ende J, Reyniers E, Fichera M, Romano C, Delle Chiaie B, Mortier G, Menten B *et al*: **Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome**. *European journal of medical genetics* 2009, **52**(2):94-100 [doi.org/doi.org/10.1016/j.ejmg.2009.02.006.](https://doi.org/https://doi.org/10.1016/j.ejmg.2009.02.006.)

152. Ramocki MB, Bartnik M, Szafranski P, Kołodziejska KE, Xia Z, Bravo J, Miller GS, Rodriguez DL, Williams CA, Bader PI *et al*: **Recurrent distal 7q11.23 deletion including HIP1 and YWHAG identified in patients with intellectual disabilities, epilepsy, and neurobehavioral problems**. *American journal of human genetics* 2010, **87**(6):857-865 [doi.org/10.1016/j.ajhg.2010.10.019.](https://doi.org/10.1016/j.ajhg.2010.10.019.)

153. Faundes V, Santa María L, Morales P, Curotto B, Parraguez MM: **Distal 7q11.23 Duplication, an Emerging Microduplication Syndrome: A Case Report and Further Characterisation**. *Mol Syndromol* 2016, **7**(5):287-291 [doi.org/10.1159/000448698.](https://doi.org/10.1159/000448698.)

154. Porter MA, Dobson-Stone C, Kwok JBJ, Schofield PR, Beckett W, Tassabehji M: **A Role for Transcription Factor GTF2IRD2 in Executive Function in Williams-Beuren Syndrome**. *PLOS ONE* 2012, **7**(10):e47457 [doi.org/10.1371/journal.pone.0047457.](https://doi.org/10.1371/journal.pone.0047457.)

155. Komoike Y, Fujii K, Nishimura A, Hiraki Y, Hayashidani M, Shimojima K, Nishizawa T, Higashi K, Yasukawa K, Saitsu H *et al*: **Zebrafish gene knockdowns imply roles for human YWHAG in infantile spasms and cardiomegaly**. *Genesis (New York, NY : 2000)* 2010, **48**(4):233-243 [doi.org/10.1002/dvg.20607.](https://doi.org/10.1002/dvg.20607.)

156. Velinov M, Ahmad A, Brown-Kipphut B, Shafiq M, Blau J, Cooma R, Roth P, Iqbal MA: **A 0.7 Mb de novo duplication at 7q21.3 including the genes DLX5 and DLX6 in a patient with split-hand/split-foot malformation**. *American journal of medical genetics Part A* 2012, **158a**(12):3201-3206 [doi.org/10.1002/ajmg.a.35644.](https://doi.org/10.1002/ajmg.a.35644.)

157. Rattanasopha S, Tongkobpetch S, Srichomthong C, Kitidumrongsook P, Suphapeetiporn K, Shotelersuk V: **Absent expression of the osteoblast-specific maternally imprinted genes, DLX5 and DLX6, causes split hand/split foot malformation type I**. *Journal of medical genetics* 2014, **51**(12):817-823 [doi.org/10.1136/jmedgenet-2014-102576.](https://doi.org/10.1136/jmedgenet-2014-102576.)

158. Tayebi N, Jamsheer A, Flöttmann R, Sowinska-Seidler A, Doelken SC, Oehl-Jaschkowitz B, Hülsemann W, Habenicht R, Klopocki E, Mundlos S *et al*: **Deletions of exons with regulatory activity at the DYNC1I1 locus are associated with split-hand/split-foot malformation: array CGH screening of 134 unrelated families**. *Orphanet J Rare Dis* 2014, **9**:108 [doi.org/10.1186/s13023-014-0108-6.](https://doi.org/10.1186/s13023-014-0108-6.)

159. Delgado S, Velinov M: **7q21.3 Deletion involving enhancer sequences within the gene DYNC1I1 presents with intellectual disability and split hand-split foot malformation with decreased penetrance**. *Molecular cytogenetics* 2015, **8**:37-37 [doi.org/10.1186/s13039-015-0139-2.](https://doi.org/10.1186/s13039-015-0139-2.)

160. Rasmussen MB, Kreiborg S, Jensen P, Bak M, Mang Y, Lodahl M, Budtz-Jørgensen E, Tommerup N, Tranebjærg L, Rendtorff ND: **Phenotypic subregions within the split-hand/foot malformation 1 locus**. *Human genetics* 2016, **135**(3):345-357 [doi.org/10.1007/s00439-016-1635-0.](https://doi.org/10.1007/s00439-016-1635-0.)

161. Lehman AM, Friedman JM, Chai D, Zahir FR, Marra MA, Prisman L, Tsang E, Eydoux P, Armstrong L: **A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7**. *European journal of medical genetics* 2009, **52**(6):436-439 [doi.org/10.1016/j.ejmg.2009.09.006.](https://doi.org/10.1016/j.ejmg.2009.09.006.)

162. Amouroux C, Vincent M, Blanchet P, Puechberty J, Schneider A, Chaze AM, Girard M, Tournaire M, Jorgensen C, Morin D *et al*: **Duplication 8q12: confirmation of a novel recognizable phenotype with duane retraction syndrome and developmental delay**. *Eur J Hum Genet* 2012, **20**(5):580-583 [doi.org/10.1038/ejhg.2011.243.](https://doi.org/10.1038/ejhg.2011.243.)

163. Luo H, Xie L, Wang SZ, Chen JL, Huang C, Wang J, Yang JF, Zhang WZ, Yang YF, Tan ZP: **Duplication of 8q12 encompassing CHD7 is associated with a distinct phenotype but without duane anomaly**. *European journal of medical genetics* 2012, **55**(11):646-649 [doi.org/10.1016/j.ejmg.2012.07.006.](https://doi.org/10.1016/j.ejmg.2012.07.006.)

164. Baroncini A, Bertuzzo S, Quarantini R, Ricciardelli P, Giorda R, Bonaglia MC: **8q12 microduplication including CHD7: clinical report on a new patient with Duane retraction syndrome type 3**. *Molecular cytogenetics* 2013, **6**(1):49-49 [doi.org/10.1186/1755-8166-6-49.](https://doi.org/10.1186/1755-8166-6-49.)

165. Palumbo O, Palumbo P, Stallone R, Palladino T, Zelante L, Carella M: **8q12.1q12.3 de novo microdeletion involving the CHD7 gene in a patient without the major features of CHARGE syndrome: Case report and critical review of the literature**. *Gene* 2013, **513**(1):209-213 [doi.org/doi.org/10.1016/j.gene.2012.09.132.](https://doi.org/https://doi.org/10.1016/j.gene.2012.09.132.)

166. Isidor B, Hamel A, Plasschaert F, Claus L, Mercier JM, Mortier GR, Leroy JG, Verloes A, David A: **Mesomelic dysplasia with acral synostoses Verloes-David-Pfeiffer type: follow-up study documents progressive clinical course**. *American journal of medical genetics Part A* 2009, **149a**(10):2220-2225 [doi.org/10.1002/ajmg.a.32926.](https://doi.org/10.1002/ajmg.a.32926.)

167. Isidor B, Pichon O, Redon R, Day-Salvatore D, Hamel A, Siwicka KA, Bitner-Glindzicz M, Heymann D, Kjellén L, Kraus C *et al*: **Mesomelia-synostoses syndrome results from deletion of SULF1 and SLCO5A1 genes at 8q13**. *American journal of human genetics* 2010, **87**(1):95-100 [doi.org/10.1016/j.ajhg.2010.05.012.](https://doi.org/10.1016/j.ajhg.2010.05.012.)

168. Hamilton MJ, Sarkar A, Dixit A, Marder E: **Phenotypes of 8q13.2-q13.3 microdeletion: Case report and literature review of an emerging recurrent microdeletion syndrome**. *American journal of medical genetics Part A* 2016, **170**(3):804-808 [doi.org/10.1002/ajmg.a.37497.](https://doi.org/10.1002/ajmg.a.37497.)

169. Palomares M, Delicado A, Mansilla E, de Torres ML, Vallespín E, Fernandez L, Martinez-Glez V, García-Miñaur S, Nevado J, Simarro FS *et al*: **Characterization of a 8q21.11 microdeletion syndrome associated with intellectual disability and a recognizable phenotype**. *American journal of human genetics* 2011, **89**(2):295-301 [doi.org/10.1016/j.ajhg.2011.06.012.](https://doi.org/10.1016/j.ajhg.2011.06.012.)

170. Niyazov D, Africk D: **Mitochondrial Dysfunction in a Patient with 8q21.11 Deletion and Charcot-Marie-Tooth Disease Type 2K due to GDAP1 Haploinsufficiency**. *Mol Syndromol* 2015, **6**(4):204-206 [doi.org/10.1159/000440660.](https://doi.org/10.1159/000440660.)

171. Quintela I, Barros F, Castro-Gago M, Carracedo A, Eiris J: **Clinical characterization of a male patient with the recently described 8q21.11 microdeletion syndrome**. *American Journal of Medical Genetics Part A* 2015, **167**(6):1369-1373 [doi.org/doi.org/10.1002/ajmg.a.37038.](https://doi.org/https://doi.org/10.1002/ajmg.a.37038.)

172. Happ H, Schilter KF, Weh E, Reis LM, Semina EV: **8q21.11 microdeletion in two patients with syndromic peters anomaly**. *American Journal of Medical Genetics Part A* 2016, **170**(9):2471-2475 [doi.org/doi.org/10.1002/ajmg.a.37840.](https://doi.org/https://doi.org/10.1002/ajmg.a.37840.)

173. Raas-Rothschild A, Dijkhuizen T, Sikkema-Raddatz B, Werner M, Dagan J, Abeliovich D, Lerer I: **The 8q22.1 microdeletion syndrome or Nablus mask-like facial syndrome: report on two patients and review of the literature**. *European journal of medical genetics* 2009, **52**(2-3):140-144 [doi.org/10.1016/j.ejmg.2009.03.011.](https://doi.org/10.1016/j.ejmg.2009.03.011.)

174. Jain S, Yang P, Farrell SA: **A case of 8q22.1 microdeletion without the Nablus mask-like facial syndrome phenotype**. *European journal of medical genetics* 2010, **53**(2):108-110 [doi.org/doi.org/10.1016/j.ejmg.2009.12.006.](https://doi.org/https://doi.org/10.1016/j.ejmg.2009.12.006.)

175. Allanson J, Smith A, Hare H, Albrecht B, Bijlsma E, Dallapiccola B, Donti E, Fitzpatrick D, Isidor B, Lachlan K *et al*: **Nablus mask-like facial syndrome: deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype**. *American journal of medical genetics Part A* 2012, **158a**(9):2091-2099 [doi.org/10.1002/ajmg.a.35446.](https://doi.org/10.1002/ajmg.a.35446.)

176. Overhoff J, Rabideau MM, Bird LM, Schweitzer DN, Haynes K, Schultz RA, Shaffer LG, Rosenfeld JA, Ellison JW: **Refinement of the 8q22.1 microdeletion critical region associated with Nablus mask-like facial syndrome**. *American Journal of Medical Genetics Part A* 2014, **164**(1):259-263 [doi.org/doi.org/10.1002/ajmg.a.36163.](https://doi.org/https://doi.org/10.1002/ajmg.a.36163.)

177. Jamuar SS, Duzkale H, Duzkale N, Zhang C, High FA, Kaban L, Bhattacharya S, Crandall B, Kantarci S, Stoler JM *et al*: **Deletion of chromosome 8q22.1, a critical region for Nablus mask-like facial syndrome: four additional cases support a role of genetic modifiers in the manifestation of the phenotype**. *American journal of medical genetics Part A* 2015, **167**(6):1400-1405 [doi.org/10.1002/ajmg.a.36848.](https://doi.org/10.1002/ajmg.a.36848.)

178. Kuechler A, Buysse K, Clayton-Smith J, Le Caignec C, David A, Engels H, Kohlhase J, Mari F, Mortier G, Renieri A *et al*: **Five patients with novel overlapping interstitial deletions in 8q22.2q22.3**. *American Journal of Medical Genetics Part A* 2011, **155**(8):1857-1864 [doi.org/doi.org/10.1002/ajmg.a.34072.](https://doi.org/https://doi.org/10.1002/ajmg.a.34072.)

179. Kuroda Y, Ohashi I, Saito T, Nagai J, Ida K, Naruto T, Iai M, Kurosawa K: **Refinement of the deletion in 8q22.2-q22.3: the minimum deletion size at 8q22.3 related to intellectual disability and epilepsy**. *American journal of medical genetics Part A* 2014, **164a**(8):2104-2108 [doi.org/10.1002/ajmg.a.36604.](https://doi.org/10.1002/ajmg.a.36604.)

180. Sinajon P, Gofine T, Ingram J, So J: **Microdeletion 8q22.2-q22.3 in a 40-year-old male**. *European journal of medical genetics* 2015, **58**(11):569-572 [doi.org/10.1016/j.ejmg.2015.10.004.](https://doi.org/10.1016/j.ejmg.2015.10.004.)

181. Pereza N, Severinski S, Ostojić S, Volk M, Maver A, Dekanić KB, Kapović M, Peterlin B: **Third case of 8q23.3-q24.13 deletion in a patient with Langer-Giedion syndrome phenotype without TRPS1 gene deletion**. *American journal of medical genetics Part A* 2012, **158a**(3):659-663 [doi.org/10.1002/ajmg.a.35201.](https://doi.org/10.1002/ajmg.a.35201.)

182. Li Q, Zhang Z, Yan Y, Xiao P, Gao Z, Cheng W, Su L, Yu K, Xie H, Chen X *et al*: **Annular pancreas in Trichorhinophalangeal syndrome type II with 8q23.3-q24.12 interstitial deletion**. *Molecular cytogenetics* 2015, **8**:95 [doi.org/10.1186/s13039-015-0201-0.](https://doi.org/10.1186/s13039-015-0201-0.)

183. Schinzel A, Riegel M, Baumer A, Superti-Furga A, Moreira LM, Santo LD, Schiper PP, Carvalho JH, Giedion A: **Long-term follow-up of four patients with Langer-Giedion syndrome: clinical course and complications**. *American journal of medical genetics Part A* 2013, **161a**(9):2216-2225 [doi.org/10.1002/ajmg.a.36062.](https://doi.org/10.1002/ajmg.a.36062.)

184. Selenti N, Tzetis M, Braoudaki M, Gianikou K, Kitsiou-Tzeli S, Fryssira H: **An interstitial deletion at 8q23.1-q24.12 associated with Langer-Giedion syndrome/ Trichorhinophalangeal syndrome (TRPS) type II and Cornelia de Lange syndrome 4**. *Molecular cytogenetics* 2015, **8**:64-64 [doi.org/10.1186/s13039-015-0169-9.](https://doi.org/10.1186/s13039-015-0169-9.)

185. Dauber A, Golzio C, Guenot C, Jodelka FM, Kibaek M, Kjaergaard S, Leheup B, Martinet D, Nowaczyk MJM, Rosenfeld JA *et al*: **SCRIB and PUF60 are primary drivers of the multisystemic phenotypes of the 8q24.3 copy-number variant**. *American journal of human genetics* 2013, **93**(5):798-811 [doi.org/10.1016/j.ajhg.2013.09.010.](https://doi.org/10.1016/j.ajhg.2013.09.010.)

186. Wells C, Spaggiari E, Malan V, Stirnemann JJ, Attie-Bitach T, Ville Y, Vekemans M, Bessieres B, Romana S: **First fetal case of the 8q24.3 contiguous genes syndrome**. *American Journal of Medical Genetics Part A* 2016, **170**(1):239-242 [doi.org/doi.org/10.1002/ajmg.a.37411.](https://doi.org/https://doi.org/10.1002/ajmg.a.37411.)

187. Abdin D, Rump A, Tzschach A, Sarnow K, Schröck E, Hackmann K, Di Donato N: **PUF60-SCRIB fusion transcript in a patient with 8q24.3 microdeletion and atypical Verheij syndrome**. *European journal of medical genetics* 2019, **62**(12):103587 [doi.org/doi.org/10.1016/j.ejmg.2018.11.021.](https://doi.org/https://doi.org/10.1016/j.ejmg.2018.11.021.)

188. Barbaro M, Balsamo A, Anderlid BM, Myhre AG, Gennari M, Nicoletti A, Pittalis MC, Oscarson M, Wedell A: **Characterization of deletions at 9p affecting the candidate regions for sex reversal and deletion 9p syndrome by MLPA**. *European Journal of Human Genetics* 2009, **17**(11):1439-1447 [doi.org/10.1038/ejhg.2009.70.](https://doi.org/10.1038/ejhg.2009.70.)

189. Tannour-Louet M, Han S, Corbett ST, Louet J-F, Yatsenko S, Meyers L, Shaw CA, Kang S-HL, Cheung SW, Lamb DJ: **Identification of de novo copy number variants associated with human disorders of sexual development**. *PloS one* 2010, **5**(10):e15392-e15392 [doi.org/10.1371/journal.pone.0015392.](https://doi.org/10.1371/journal.pone.0015392.)

190. Onesimo R, Orteschi D, Scalzone M, Rossodivita A, Nanni L, Zannoni GF, Marrocco G, Battaglia D, Fundarò C, Neri G: **Chromosome 9p deletion syndrome and sex reversal: Novel findings and redefinition of the critically deleted regions**. *American Journal of Medical Genetics Part A* 2012, **158A**(9):2266-2271 [doi.org/doi.org/10.1002/ajmg.a.35489.](https://doi.org/https://doi.org/10.1002/ajmg.a.35489.)

191. Quinonez SC, Park JM, Rabah R, Owens KM, Yashar BM, Glover TW, Keegan CE: **9p partial monosomy and disorders of sex development: Review and postulation of a pathogenetic mechanism**. *American Journal of Medical Genetics Part A* 2013, **161**(8):1882-1896 [doi.org/doi.org/10.1002/ajmg.a.36018.](https://doi.org/https://doi.org/10.1002/ajmg.a.36018.)

192. Hauge X, Raca G, Cooper S, May K, Spiro R, Adam M, Martin CL: **Detailed characterization of, and clinical correlations in, 10 patients with distal deletions of chromosome 9p**. *Genetics in Medicine* 2008, **10**(8):599-611 [doi.org/10.1097/GIM.0b013e31817e2bde.](https://doi.org/10.1097/GIM.0b013e31817e2bde.)

193. Swinkels ME, Simons A, Smeets DF, Vissers LE, Veltman JA, Pfundt R, de Vries BB, Faas BH, Schrander-Stumpel CT, McCann E *et al*: **Clinical and cytogenetic characterization of 13 Dutch patients with deletion 9p syndrome: Delineation of the critical region for a consensus phenotype**. *American journal of medical genetics Part A* 2008, **146a**(11):1430-1438 [doi.org/10.1002/ajmg.a.32310.](https://doi.org/10.1002/ajmg.a.32310.)

194. Mitsui N, Shimizu K, Nishimoto H, Mochizuki H, Iida M, Ohashi H: **Patient with terminal 9 Mb deletion of chromosome 9p: Refining the critical region for 9p monosomy syndrome with trigonocephaly**. *Congenital anomalies* 2013, **53**(1):49-53 [doi.org/doi.org/10.1111/j.1741-4520.2012.00362.x.](https://doi.org/https://doi.org/10.1111/j.1741-4520.2012.00362.x.)

195. Niemi AK, Kwan A, Hudgins L, Cherry AM, Manning MA: **Report of two patients and further characterization of interstitial 9p13 deletion--a rare but recurrent microdeletion syndrome?** *American journal of medical genetics Part A* 2012, **158a**(9):2328-2335 [doi.org/10.1002/ajmg.a.35536.](https://doi.org/10.1002/ajmg.a.35536.)

196. Crone M, Thomas MA: **9p13.1p13.3 interstitial deletion: A case report and further delineation of a rare condition**. *American Journal of Medical Genetics Part A* 2016, **170**(4):1095-1098 [doi.org/doi.org/10.1002/ajmg.a.37534.](https://doi.org/https://doi.org/10.1002/ajmg.a.37534.)

197. Ferreira SI, Cinnirella G, Ramos L, Suppa A, Pires LM, Nardone AM, Camerota L, Lanciotti S, Galasso C, De Maio F *et al*: **Tremor is a major feature of 9p13 deletion syndrome**. *American Journal of Medical Genetics Part A* 2020, **182**(11):2694-2698 [doi.org/doi.org/10.1002/ajmg.a.61807.](https://doi.org/https://doi.org/10.1002/ajmg.a.61807.)

198. Shimojima K, Adachi M, Tanaka M, Tanaka Y, Kurosawa K, Yamamoto T: **Clinical features of microdeletion 9q22.3 (pat)**. *Clin Genet* 2009, **75**(4):384-393 [doi.org/10.1111/j.1399-0004.2008.01141.x.](https://doi.org/10.1111/j.1399-0004.2008.01141.x.)

199. Muller EA, Aradhya S, Atkin JF, Carmany EP, Elliott AM, Chudley AE, Clark RD, Everman DB, Garner S, Hall BD *et al*: **Microdeletion 9q22.3 syndrome includes metopic craniosynostosis, hydrocephalus, macrosomia, and developmental delay**. *American journal of medical genetics Part A* 2012, **158a**(2):391-399 [doi.org/10.1002/ajmg.a.34216.](https://doi.org/10.1002/ajmg.a.34216.)

200. Reichert SC, Zelley K, Nichols KE, Eberhard M, Zackai EH, Martinez-Poyer J: **Diagnosis of 9q22.3 microdeletion syndrome in utero following identification of craniosynostosis, overgrowth, and skeletal anomalies**. *American journal of medical genetics Part A* 2015, **167a**(4):862-865 [doi.org/10.1002/ajmg.a.37013.](https://doi.org/10.1002/ajmg.a.37013.)

201. Yamada H, Shimura M, Takahashi H, Nara S, Morishima Y, Go S, Miyashita T, Numabe H, Kawashima H: **A familial case of overgrowth syndrome caused by a 9q22.3 microdeletion in a mother and daughter**. *European journal of medical genetics* 2020, **63**(5):103872 [doi.org/10.1016/j.ejmg.2020.103872.](https://doi.org/10.1016/j.ejmg.2020.103872.)

202. Ewing AD, Cheetham SW, McGill JJ, Sharkey M, Walker R, West JA, West MJ, Summers KM: **Microdeletion of 9q22.3: A patient with minimal deletion size associated with a severe phenotype**. *American Journal of Medical Genetics Part A* 2021, **n/a**(n/a) [doi.org/doi.org/10.1002/ajmg.a.62224.](https://doi.org/https://doi.org/10.1002/ajmg.a.62224.)

203. Mucciolo M, Magini P, Marozza A, Mongelli P, Mencarelli MA, Hayek G, Tavalazzi F, Mari F, Seri M, Renieri A *et al*: **9q31.1q31.3 deletion in two patients with similar clinical features: a newly recognized microdeletion syndrome?** *American journal of medical genetics Part A* 2014, **164a**(3):685-690 [doi.org/10.1002/ajmg.a.36361.](https://doi.org/10.1002/ajmg.a.36361.)

204. Nevado J, Mergener R, Palomares-Bralo M, Souza KR, Vallespín E, Mena R, Martínez-Glez V, Mori M, Santos F, García-Miñaur S *et al*: **New microdeletion and microduplication syndromes: A comprehensive review**. *Genetics and molecular biology* 2014, **37**(1 Suppl):210-219 [doi.org/10.1590/s1415-47572014000200007.](https://doi.org/10.1590/s1415-47572014000200007.)

205. Schuffenhauer S, Lichtner P, Peykar-Derakhshandeh P, Murken J, Haas OA, Back E, Wolff G, Zabel B, Barisic I, Rauch A *et al*: **Deletion mapping on chromosome 10p and definition of a critical region for the second DiGeorge syndrome locus (DGS2)**. *Eur J Hum Genet* 1998, **6**(3):213-225 [doi.org/10.1038/sj.ejhg.5200183.](https://doi.org/10.1038/sj.ejhg.5200183.)

206. Lichtner P, König R, Hasegawa T, Van Esch H, Meitinger T, Schuffenhauer S: **An HDR (hypoparathyroidism, deafness, renal dysplasia) syndrome locus maps distal to the DiGeorge syndrome region on 10p13/14**. *Journal of medical genetics* 2000, **37**(1):33-37 [doi.org/10.1136/jmg.37.1.33.](https://doi.org/10.1136/jmg.37.1.33.)

207. Skrypnyk C, Goecke TO, Majewski F, Bartsch O: **Molecular cytogenetic characterization of a 10p14 deletion that includes the DGS2 region in a patient with multiple anomalies**. *American journal of medical genetics* 2002, **113**(2):207-212 [doi.org/10.1002/ajmg.10764.](https://doi.org/10.1002/ajmg.10764.)

208. Yatsenko SA, Yatsenko AN, Szigeti K, Craigen WJ, Stankiewicz P, Cheung SW, Lupski JR: **Interstitial deletion of 10p and atrial septal defect in DiGeorge 2 syndrome**. *Clin Genet* 2004, **66**(2):128-136 [doi.org/10.1111/j.1399-0004.2004.00290.x.](https://doi.org/10.1111/j.1399-0004.2004.00290.x.)

209. Lindstrand A, Malmgren H, Verri A, Benetti E, Eriksson M, Nordgren A, Anderlid BM, Golovleva I, Schoumans J, Blennow E: **Molecular and clinical characterization of patients with overlapping 10p deletions**. *American journal of medical genetics Part A* 2010, **152a**(5):1233-1243 [doi.org/10.1002/ajmg.a.33366.](https://doi.org/10.1002/ajmg.a.33366.)

210. Melis D, Genesio R, Boemio P, Del Giudice E, Cappuccio G, Mormile A, Ronga V, Conti A, Imperati F, Nitsch L *et al*: **Clinical description of a patient carrying the smallest reported deletion involving 10p14 region**. *American Journal of Medical Genetics Part A* 2012, **158A**(4):832-835 [doi.org/doi.org/10.1002/ajmg.a.34133.](https://doi.org/https://doi.org/10.1002/ajmg.a.34133.)

211. Kim SB, Kim Y-E, Jung JM, Jin HY, Lim Y-J, Chung ML: **Clinical description of a neonate carrying the largest reported deletion involving the 10p15.3p13 region**. *Clin Case Rep* 2017, **5**(8):1369-1375 [doi.org/10.1002/ccr3.1070.](https://doi.org/10.1002/ccr3.1070.)

212. Shao Q-Y, Wu P-L, Lin B-Y, Chen S-J, Liu J, Chen S-Q: **Clinical report of a neonate carrying a large deletion in the 10p15.3p13 region and review of the literature**. *Molecular cytogenetics* 2021, **14**(1):29 [doi.org/10.1186/s13039-021-00546-1.](https://doi.org/10.1186/s13039-021-00546-1.)

213. DeScipio C, Conlin L, Rosenfeld J, Tepperberg J, Pasion R, Patel A, McDonald MT, Aradhya S, Ho D, Goldstein J *et al*: **Subtelomeric deletion of chromosome 10p15.3: Clinical findings and molecular cytogenetic characterization**. *American Journal of Medical Genetics Part A* 2012, **158A**(9):2152-2161 [doi.org/doi.org/10.1002/ajmg.a.35574.](https://doi.org/https://doi.org/10.1002/ajmg.a.35574.)

214. Poluha A, Bernaciak J, Jaszczuk I, Kędzior M, Nowakowska BA: **Molecular and clinical characterization of new patient with 1,08 Mb deletion in 10p15.3 region**. *Molecular cytogenetics* 2017, **10**(1):34 [doi.org/10.1186/s13039-017-0336-2.](https://doi.org/10.1186/s13039-017-0336-2.)

215. Van Esch H, Groenen P, Nesbit MA, Schuffenhauer S, Lichtner P, Vanderlinden G, Harding B, Beetz R, Bilous RW, Holdaway I *et al*: **GATA3 haplo-insufficiency causes human HDR syndrome**. *Nature* 2000, **406**(6794):419-422 [doi.org/10.1038/35019088.](https://doi.org/10.1038/35019088.)

216. Fukami M, Muroya K, Miyake T, Iso M, Kato F, Yokoi H, Suzuki Y, Tsubouchi K, Nakagomi Y, Kikuchi N *et al*: **<i>GATA3</i> abnormalities in six patients with HDR syndrome**. *Endocrine Journal* 2011, **58**(2):117-121 [doi.org/10.1507/endocrj.K10E-234.](https://doi.org/10.1507/endocrj.K10E-234.)

217. Lichtner P, Attié-Bitach T, Schuffenhauer S, Henwood J, Bouvagnet P, Scambler PJ, Meitinger T, Vekemans M: **Expression and mutation analysis of BRUNOL3, a candidate gene for heart and thymus developmental defects associated with partial monosomy 10p**. *Journal of molecular medicine (Berlin, Germany)* 2002, **80**(7):431-442 [doi.org/10.1007/s00109-002-0331-9.](https://doi.org/10.1007/s00109-002-0331-9.)

218. Alliman S, Coppinger J, Marcadier J, Thiese H, Brock P, Shafer S, Weaver C, Asamoah A, Leppig K, Dyack S *et al*: **Clinical and molecular characterization of individuals with recurrent genomic disorder at 10q22.3q23.2**. *Clin Genet* 2010, **78**(2):162-168 [doi.org/10.1111/j.1399-0004.2010.01373.x.](https://doi.org/10.1111/j.1399-0004.2010.01373.x.)

219. van Bon BW, Balciuniene J, Fruhman G, Nagamani SC, Broome DL, Cameron E, Martinet D, Roulet E, Jacquemont S, Beckmann JS *et al*: **The phenotype of recurrent 10q22q23 deletions and duplications**. *Eur J Hum Genet* 2011, **19**(4):400-408 [doi.org/10.1038/ejhg.2010.211.](https://doi.org/10.1038/ejhg.2010.211.)

220. Dimitrov BI, de Ravel T, Van Driessche J, de Die-Smulders C, Toutain A, Vermeesch JR, Fryns JP, Devriendt K, Debeer P: **Distal limb deficiencies, micrognathia syndrome, and syndromic forms of split hand foot malformation (SHFM) are caused by chromosome 10q genomic rearrangements**. *Journal of medical genetics* 2010, **47**(2):103-111 [doi.org/10.1136/jmg.2008.065888.](https://doi.org/10.1136/jmg.2008.065888.)

221. Holder-Espinasse M, Jamsheer A, Escande F, Andrieux J, Petit F, Sowinska-Seidler A, Socha M, Jakubiuk-Tomaszuk A, Gerard M, Mathieu-Dramard M *et al*: **Duplication of 10q24 locus: broadening the clinical and radiological spectrum**. *European Journal of Human Genetics* 2019, **27**(4):525-534 [doi.org/10.1038/s41431-018-0326-9.](https://doi.org/10.1038/s41431-018-0326-9.)

222. Choucair N, Abou Ghoch J, Fawaz A, Mégarbané A, Chouery E: **10q26.1 Microdeletion: Redefining the critical regions for microcephaly and genital anomalies**. *American journal of medical genetics Part A* 2015, **167a**(11):2707-2713 [doi.org/10.1002/ajmg.a.37211.](https://doi.org/10.1002/ajmg.a.37211.)

223. Faria ÁC, Rabbi-Bortolini E, Rebouças MRGO, de S. Thiago Pereira ALA, Frasson MGT, Atique R, Lourenço NCV, Rosenberg C, Kobayashi GS, Passos-Bueno MR *et al*: **Craniosynostosis in 10q26 deletion patients: A consequence of brain underdevelopment or altered suture biology?** *American Journal of Medical Genetics Part A* 2016, **170**(2):403-409 [doi.org/doi.org/10.1002/ajmg.a.37448.](https://doi.org/https://doi.org/10.1002/ajmg.a.37448.)

224. Lin S, Zhou Y, Fang Q, Wu J, Zhang Z, Ji Y, Luo Y: **Chromosome 10q26 deletion syndrome: Two new cases and a review of the literature**. *Molecular medicine reports* 2016, **14**(6):5134-5140 [doi.org/10.3892/mmr.2016.5864.](https://doi.org/10.3892/mmr.2016.5864.)

225. Yatsenko SA, Kruer MC, Bader PI, Corzo D, Schuette J, Keegan CE, Nowakowska B, Peacock S, Cai WW, Peiffer DA *et al*: **Identification of critical regions for clinical features of distal 10q deletion syndrome**. *Clinical Genetics* 2009, **76**(1):54-62 [doi.org/doi.org/10.1111/j.1399-0004.2008.01115.x.](https://doi.org/https://doi.org/10.1111/j.1399-0004.2008.01115.x.)

226. Chiesa N, De Crescenzo A, Mishra K, Perone L, Carella M, Palumbo O, Mussa A, Sparago A, Cerrato F, Russo S *et al*: **The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases**. *Hum Mol Genet* 2012, **21**(1):10-25 [doi.org/10.1093/hmg/ddr419.](https://doi.org/10.1093/hmg/ddr419.)

227. Addis L, Ahn JW, Dobson R, Dixit A, Ogilvie CM, Pinto D, Vaags AK, Coon H, Chaste P, Wilson S *et al*: **Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation**. *Human mutation* 2015, **36**(9):842-850 [doi.org/10.1002/humu.22816.](https://doi.org/10.1002/humu.22816.)

228. Han JC, Liu Q-R, Jones M, Levinn RL, Menzie CM, Jefferson-George KS, Adler-Wailes DC, Sanford EL, Lacbawan FL, Uhl GR *et al*: **Brain-Derived Neurotrophic Factor and Obesity in the WAGR Syndrome**. *New England Journal of Medicine* 2008, **359**(9):918-927 [doi.org/10.1056/NEJMoa0801119.](https://doi.org/10.1056/NEJMoa0801119.)

229. Han JC, Thurm A, Golden Williams C, Joseph LA, Zein WM, Brooks BP, Butman JA, Brady SM, Fuhr SR, Hicks MD *et al*: **Association of brain-derived neurotrophic factor (BDNF) haploinsufficiency with lower adaptive behaviour and reduced cognitive functioning in WAGR/11p13 deletion syndrome**. *Cortex* 2013, **49**(10):2700-2710 [doi.org/doi.org/10.1016/j.cortex.2013.02.009.](https://doi.org/https://doi.org/10.1016/j.cortex.2013.02.009.)

230. Ferreira MAT, Almeida Júnior IG, Kuratani DK, Rosa RFM, Gonzales JFO, Telles LEB, Ferrão YA, Zen PRG: **WAGRO syndrome: a rare genetic condition associated with aniridia and additional ophthalmologic abnormalities**. *Arquivos brasileiros de oftalmologia* 2019, **82**(4):336-338 [doi.org/10.5935/0004-2749.20190065.](https://doi.org/10.5935/0004-2749.20190065.)

231. Hall HN, Williamson KA, FitzPatrick DR: **The genetic architecture of aniridia and Gillespie syndrome**. *Human genetics* 2019, **138**(8-9):881-898 [doi.org/10.1007/s00439-018-1934-8.](https://doi.org/10.1007/s00439-018-1934-8.)

232. Fischbach BV, Trout KL, Lewis J, Luis CA, Sika M: **WAGR Syndrome: A Clinical Review of 54 Cases**. *Pediatrics* 2005, **116**(4):984 [doi.org/10.1542/peds.2004-0467.](https://doi.org/10.1542/peds.2004-0467.)

233. Yamamoto T, Togawa M, Shimada S, Sangu N, Shimojima K, Okamoto N: **Narrowing of the responsible region for severe developmental delay and autistic behaviors in WAGR syndrome down to 1.6 Mb including PAX6, WT1, and PRRG4**. *American journal of medical genetics Part A* 2014, **164a**(3):634-638 [doi.org/10.1002/ajmg.a.36325.](https://doi.org/10.1002/ajmg.a.36325.)

234. Swarr DT, Bloom D, Lewis RA, Elenberg E, Friedman EM, Glotzbach C, Wissman SD, Shaffer LG, Potocki L: **Potocki-Shaffer syndrome: comprehensive clinical assessment, review of the literature, and proposals for medical management**. *American journal of medical genetics Part A* 2010, **152a**(3):565-572 [doi.org/10.1002/ajmg.a.33245.](https://doi.org/10.1002/ajmg.a.33245.)

235. Kim H-G, Kim H-T, Leach NT, Lan F, Ullmann R, Silahtaroglu A, Kurth I, Nowka A, Seong IS, Shen Y *et al*: **Translocations disrupting PHF21A in the Potocki-Shaffer-syndrome region are associated with intellectual disability and craniofacial anomalies**. *American journal of human genetics* 2012, **91**(1):56-72 [doi.org/10.1016/j.ajhg.2012.05.005.](https://doi.org/10.1016/j.ajhg.2012.05.005.)

236. Kim HG, Rosenfeld JA, Scott DA, Bénédicte G, Labonne JD, Brown J, McGuire M, Mahida S, Naidu S, Gutierrez J *et al*: **Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism**. *Molecular autism* 2019, **10**:35 [doi.org/10.1186/s13229-019-0286-0.](https://doi.org/10.1186/s13229-019-0286-0.)

237. Trajkova S, Di Gregorio E, Ferrero GB, Carli D, Pavinato L, Delplancq G, Kuentz P, Brusco A: **New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review**. *Brain sciences* 2020, **10**(11) [doi.org/10.3390/brainsci10110788.](https://doi.org/10.3390/brainsci10110788.)

238. Gregory-Evans CY, Moosajee M, Hodges MD, Mackay DS, Game L, Vargesson N, Bloch-Zupan A, Rüschendorf F, Santos-Pinto L, Wackens G *et al*: **SNP genome scanning localizes oto-dental syndrome to chromosome 11q13 and microdeletions at this locus implicate FGF3 in dental and inner-ear disease and FADD in ocular coloboma**. *Hum Mol Genet* 2007, **16**(20):2482-2493 [doi.org/10.1093/hmg/ddm204.](https://doi.org/10.1093/hmg/ddm204.)

239. Alsmadi O, Meyer BF, Alkuraya F, Wakil S, Alkayal F, Al-Saud H, Ramzan K, Al-Sayed M: **Syndromic congenital sensorineural deafness, microtia and microdontia resulting from a novel homoallelic mutation in fibroblast growth factor 3 (FGF3)**. *European Journal of Human Genetics* 2009, **17**(1):14-21 [doi.org/10.1038/ejhg.2008.141.](https://doi.org/10.1038/ejhg.2008.141.)

240. Ziebart T, Draenert FG, Galetzka D, Babaryka G, Schmidseder R, Wagner W, Bartsch O: **The original family revisited after 37 years: odontoma–dysphagia syndrome is most likely caused by a microduplication of chromosome 11q13.3, including the FGF3 and FGF4 genes**. *Clinical Oral Investigations* 2013, **17**(1):123-130 [doi.org/10.1007/s00784-012-0676-6.](https://doi.org/10.1007/s00784-012-0676-6.)

241. Kim Y-S, Kim G-H, Byeon JH, Eun S-H, Eun B-L: **Chromosome 11q13 deletion syndrome**. *Korean J Pediatr* 2016, **59**(Suppl 1):S10-S13 [doi.org/10.3345/kjp.2016.59.11.S10.](https://doi.org/10.3345/kjp.2016.59.11.S10.)

242. Grossfeld PD, Mattina T, Lai Z, Favier R, Jones KL, Cotter F, Jones C, the 11q C: **The 11q terminal deletion disorder: A prospective study of 110 cases**. *American Journal of Medical Genetics Part A* 2004, **129A**(1):51-61 [doi.org/doi.org/10.1002/ajmg.a.30090.](https://doi.org/https://doi.org/10.1002/ajmg.a.30090.)

243. Coldren CD, Lai Z, Shragg P, Rossi E, Glidewell SC, Zuffardi O, Mattina T, Ivy DD, Curfs LM, Mattson SN *et al*: **Chromosomal microarray mapping suggests a role for BSX and Neurogranin in neurocognitive and behavioral defects in the 11q terminal deletion disorder (Jacobsen syndrome)**. *neurogenetics* 2008, **10**(2):89 [doi.org/10.1007/s10048-008-0157-x.](https://doi.org/10.1007/s10048-008-0157-x.)

244. Ji T, Wu Y, Wang H, Wang J, Jiang Y: **Diagnosis and fine mapping of a deletion in distal 11q in two Chinese patients with developmental delay**. *Journal of Human Genetics* 2010, **55**(8):486-489 [doi.org/10.1038/jhg.2010.51.](https://doi.org/10.1038/jhg.2010.51.)

245. Ye M, Coldren C, Liang X, Mattina T, Goldmuntz E, Benson DW, Ivy D, Perryman MB, Garrett-Sinha LA, Grossfeld P: **Deletion of ETS-1, a gene in the Jacobsen syndrome critical region, causes ventricular septal defects and abnormal ventricular morphology in mice**. *Hum Mol Genet* 2010, **19**(4):648-656 [doi.org/10.1093/hmg/ddp532.](https://doi.org/10.1093/hmg/ddp532.)

246. Rooryck C, Stef M, Burgelin I, Simon D, Souakri N, Thambo JB, Chateil JF, Lacombe D, Arveiler B: **2.3 Mb terminal deletion in 12p13.33 associated with oculoauriculovertebral spectrum and evaluation of WNT5B as a candidate gene**. *European journal of medical genetics* 2009, **52**(6):446-449 [doi.org/10.1016/j.ejmg.2009.08.005.](https://doi.org/10.1016/j.ejmg.2009.08.005.)

247. Thevenon J, Callier P, Andrieux J, Delobel B, David A, Sukno S, Minot D, Mosca Anne L, Marle N, Sanlaville D *et al*: **12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech**. *Eur J Hum Genet* 2013, **21**(1):82-88 [doi.org/10.1038/ejhg.2012.116.](https://doi.org/10.1038/ejhg.2012.116.)

248. Silva IM, Rosenfeld J, Antoniuk SA, Raskin S, Sotomaior VS: **A 1.5Mb terminal deletion of 12p associated with autism spectrum disorder**. *Gene* 2014, **542**(1):83-86 [doi.org/10.1016/j.gene.2014.02.058.](https://doi.org/10.1016/j.gene.2014.02.058.)

249. Mio C, Passon N, Baldan F, Bregant E, Monaco E, Mancini L, Demori E, Damante G: **CACNA1C haploinsufficiency accounts for the common features of interstitial 12p13.33 deletion carriers**. *European journal of medical genetics* 2020, **63**(4):103843 [doi.org/10.1016/j.ejmg.2020.103843.](https://doi.org/10.1016/j.ejmg.2020.103843.)

250. Menten B, Buysse K, Zahir F, Hellemans J, Hamilton SJ, Costa T, Fagerstrom C, Anadiotis G, Kingsbury D, McGillivray BC *et al*: **Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14**. *Journal of medical genetics* 2007, **44**(4):264-268 [doi.org/10.1136/jmg.2006.047860.](https://doi.org/10.1136/jmg.2006.047860.)

251. Buysse K, Reardon W, Mehta L, Costa T, Fagerstrom C, Kingsbury DJ, Anadiotis G, McGillivray BC, Hellemans J, de Leeuw N *et al*: **The 12q14 microdeletion syndrome: additional patients and further evidence that HMGA2 is an important genetic determinant for human height**. *European journal of medical genetics* 2009, **52**(2-3):101-107 [doi.org/10.1016/j.ejmg.2009.03.001.](https://doi.org/10.1016/j.ejmg.2009.03.001.)

252. Lynch SA, Foulds N, Thuresson AC, Collins AL, Annerén G, Hedberg BO, Delaney CA, Iremonger J, Murray CM, Crolla JA *et al*: **The 12q14 microdeletion syndrome: six new cases confirming the role of HMGA2 in growth**. *Eur J Hum Genet* 2011, **19**(5):534-539 [doi.org/10.1038/ejhg.2010.215.](https://doi.org/10.1038/ejhg.2010.215.)

253. Fischetto R, Palumbo O, Ortolani F, Palumbo P, Leone MP, Causio FA, Pesce S, Digilio MC, Carella M, Papadia F: **Clinical and molecular characterization of a second family with the 12q14 microdeletion syndrome and review of the literature**. *American Journal of Medical Genetics Part A* 2017, **173**(7):1922-1930 [doi.org/doi.org/10.1002/ajmg.a.38253.](https://doi.org/https://doi.org/10.1002/ajmg.a.38253.)

254. Dória S, Alves D, Pinho MJ, Pinto J, Leão M: **12q14 microduplication: a new clinical entity reciprocal to the microdeletion syndrome?** *BMC Medical Genomics* 2020, **13**(1):2 [doi.org/10.1186/s12920-019-0653-x.](https://doi.org/10.1186/s12920-019-0653-x.)

255. Aldave AJ, Rosenwasser GO, Yellore VS, Papp JC, Sobel EM, Pham MN, Chen MC, Dandekar S, Sripracha R, Rayner SA *et al*: **Linkage of posterior amorphous corneal dystrophy to chromosome 12q21.33 and exclusion of coding region mutations in KERA, LUM, DCN, and EPYC**. *Investigative ophthalmology & visual science* 2010, **51**(8):4006-4012 [doi.org/10.1167/iovs.09-4067.](https://doi.org/10.1167/iovs.09-4067.)

256. Kim MJ, Frausto RF, Rosenwasser GO, Bui T, Le DJ, Stone EM, Aldave AJ: **Posterior amorphous corneal dystrophy is associated with a deletion of small leucine-rich proteoglycans on chromosome 12**. *PLoS One* 2014, **9**(4):e95037 [doi.org/10.1371/journal.pone.0095037.](https://doi.org/10.1371/journal.pone.0095037.)

257. Odent S, Casteels I, Cassiman C, Dieltiëns M, Hua MT, Devriendt K: **Posterior amorphous corneal dystrophy caused by a de novo deletion**. *Ophthalmic genetics* 2017, **38**(2):167-170 [doi.org/10.3109/13816810.2016.1164194.](https://doi.org/10.3109/13816810.2016.1164194.)

258. Baple E, Palmer R, Hennekam RCM: **A microdeletion at 12q24.31 can mimic beckwith-wiedemann syndrome neonatally**. *Molecular syndromology* 2010, **1**(1):42-45 [doi.org/10.1159/000275671.](https://doi.org/10.1159/000275671.)

259. Qiao Y, Tyson C, Hrynchak M, Lopez-Rangel E, Hildebrand J, Martell S, Fawcett C, Kasmara L, Calli K, Harvard C *et al*: **Clinical application of 2.7M Cytogenetics array for CNV detection in subjects with idiopathic autism and/or intellectual disability**. *Clin Genet* 2013, **83**(2):145-154 [doi.org/10.1111/j.1399-0004.2012.01860.x.](https://doi.org/10.1111/j.1399-0004.2012.01860.x.)

260. Chouery E, Choucair N, Abou Ghoch J, El Sabbagh S, Corbani S, Mégarbané A: **Report on a patient with a 12q24.31 microdeletion inherited from an insulin-dependent diabetes mellitus father**. *Molecular syndromology* 2013, **4**(3):136-142 [doi.org/10.1159/000346473.](https://doi.org/10.1159/000346473.)

261. Palumbo O, Palumbo P, Delvecchio M, Palladino T, Stallone R, Crisetti M, Zelante L, Carella M: **Microdeletion of 12q24.31: report of a girl with intellectual disability, stereotypies, seizures and facial dysmorphisms**. *American journal of medical genetics Part A* 2015, **167a**(2):438-444 [doi.org/10.1002/ajmg.a.36872.](https://doi.org/10.1002/ajmg.a.36872.)

262. Labonne JDJ, Lee K-H, Iwase S, Kong I-K, Diamond MP, Layman LC, Kim C-H, Kim H-G: **An atypical 12q24.31 microdeletion implicates six genes including a histone demethylase KDM2B and a histone methyltransferase SETD1B in syndromic intellectual disability**. *Human genetics* 2016, **135**(7):757-771 [doi.org/10.1007/s00439-016-1668-4.](https://doi.org/10.1007/s00439-016-1668-4.)

263. Der Kaloustian VM, Russell L, Aradhya S, Richard G, Rosenblatt B, Melançon S: **A de novo 2.1-Mb deletion of 13q12.11 in a child with developmental delay and minor dysmorphic features**. *American journal of medical genetics Part A* 2011, **155a**(10):2538-2542 [doi.org/10.1002/ajmg.a.34198.](https://doi.org/10.1002/ajmg.a.34198.)

264. Tanteles GA, Dixit A, Smith N, Martin K, Suri M: **Mild phenotype in a patient with a de-novo 2.9-Mb interstitial deletion at 13q12.11**. *Clinical dysmorphology* 2011, **20**(2):61-65 [doi.org/10.1097/MCD.0b013e3283448498.](https://doi.org/10.1097/MCD.0b013e3283448498.)

265. Lagou M, Papoulidis I, Orru S, Papadopoulos V, Daskalakis G, Kontodiou M, Anastasakis E, Petersen MB, Kitsos G, Thomaidis L *et al*: **A de novo 2.9 Mb interstitial deletion at 13q12.11 in a child with developmental delay accompanied by mild dysmorphic characteristics**. *Molecular cytogenetics* 2014, **7**(1):92-92 [doi.org/10.1186/s13039-014-0092-5.](https://doi.org/10.1186/s13039-014-0092-5.)

266. Pavone P, Briuglia S, Falsaperla R, Warm A, Pavone V, Bernardini L, Novelli A, Praticò AD, Salpietro V, Ruggieri M: **Wide spectrum of congenital anomalies including choanal atresia, malformed extremities, and brain and spinal malformations in a girl with a de novo 5.6-Mb deletion of 13q12.11-13q12.13**. *American journal of medical genetics Part A* 2014, **164a**(7):1734-1743 [doi.org/10.1002/ajmg.a.36391.](https://doi.org/10.1002/ajmg.a.36391.)

267. Tominaga M, Saito T, Masuno M, Umeda Y, Kurosawa K: **Developmental delay and dysmorphic features in a girl with a de novo 5.4 Mb deletion of 13q12.11-q12.13**. *Congenital anomalies* 2019 [doi.org/10.1111/cga.12346.](https://doi.org/10.1111/cga.12346.)

268. Bartholdi D, Stray-Pedersen A, Azzarello-Burri S, Kibaek M, Kirchhoff M, Oneda B, Rødningen O, Schmitt-Mechelke T, Rauch A, Kjaergaard S: **A newly recognized 13q12.3 microdeletion syndrome characterized by intellectual disability, microcephaly, and eczema/atopic dermatitis encompassing the HMGB1 and KATNAL1 genes**. *American journal of medical genetics Part A* 2014, **164a**(5):1277-1283 [doi.org/10.1002/ajmg.a.36439.](https://doi.org/10.1002/ajmg.a.36439.)

269. Mandrile G, Di Gregorio E, Calcia A, Brussino A, Grosso E, Savin E, Giachino DF, Brusco A: **A New Case of 13q12.2q13.1 Microdeletion Syndrome Contributes to Phenotype Delineation**. *Case Reports in Genetics* 2014, **2014**:470830 [doi.org/10.1155/2014/470830.](https://doi.org/10.1155/2014/470830.)

270. Wang M, Li B, Liao Z, Jia Y, Fu Y: **A novel phenotype of 13q12.3 microdeletion characterized by epilepsy in an Asian child: a case report**. *BMC Medical Genomics* 2020, **13**(1):144 [doi.org/10.1186/s12920-020-00801-1.](https://doi.org/10.1186/s12920-020-00801-1.)

271. Uguen K, Krysiak K, Audebert-Bellanger S, Redon S, Benech C, Viora-Dupont E, Tran Mau-Them F, Rondeau S, Elsharkawi I, Granadillo JL *et al*: **Heterozygous HMGB1 loss-of-function variants are associated with developmental delay and microcephaly**. *Clinical Genetics* 2021, **n/a**(n/a) [doi.org/doi.org/10.1111/cge.14015.](https://doi.org/https://doi.org/10.1111/cge.14015.)

272. Caselli R, Speciale C, Pescucci C, Uliana V, Sampieri K, Bruttini M, Longo I, De Francesco S, Pramparo T, Zuffardi O *et al*: **Retinoblastoma and mental retardation microdeletion syndrome: clinical characterization and molecular dissection using array CGH**. *J Hum Genet* 2007, **52**(6):535-542 [doi.org/10.1007/s10038-007-0151-4.](https://doi.org/10.1007/s10038-007-0151-4.)

273. Mitter D, Ullmann R, Muradyan A, Klein-Hitpass L, Kanber D, Ounap K, Kaulisch M, Lohmann D: **Genotype-phenotype correlations in patients with retinoblastoma and interstitial 13q deletions**. *Eur J Hum Genet* 2011, **19**(9):947-958 [doi.org/10.1038/ejhg.2011.58.](https://doi.org/10.1038/ejhg.2011.58.)

274. Castéra L, Dehainault C, Michaux D, Lumbroso-Le Rouic L, Aerts I, Doz F, Pelet A, Couturier J, Stoppa-Lyonnet D, Gauthier-Villars M *et al*: **Fine mapping of whole RB1 gene deletions in retinoblastoma patients confirms PCDH8 as a candidate gene for psychomotor delay**. *European Journal of Human Genetics* 2013, **21**(4):460-464 [doi.org/10.1038/ejhg.2012.186.](https://doi.org/10.1038/ejhg.2012.186.)

275. de Pontual L, Yao E, Callier P, Faivre L, Drouin V, Cariou S, Van Haeringen A, Geneviève D, Goldenberg A, Oufadem M *et al*: **Germline deletion of the miR-17∼92 cluster causes skeletal and growth defects in humans**. *Nature genetics* 2011, **43**(10):1026-1030 [doi.org/10.1038/ng.915.](https://doi.org/10.1038/ng.915.)

276. Ganjavi H, Siu VM, Speevak M, MacDonald PA: **A fourth case of Feingold syndrome type 2: psychiatric presentation and management**. *BMJ case reports* 2014, **2014** [doi.org/10.1136/bcr-2014-207501.](https://doi.org/10.1136/bcr-2014-207501.)

277. Valdes-Miranda JM, Soto-Alvarez JR, Toral-Lopez J, González-Huerta L, Perez-Cabrera A, Gonzalez-Monfil G, Messina-Bass O, Cuevas-Covarrubias S: **A novel microdeletion involving the 13q31.3-q32.1 region in a patient with normal intelligence**. *European journal of medical genetics* 2014, **57**(2-3):60-64 [doi.org/10.1016/j.ejmg.2014.01.006.](https://doi.org/10.1016/j.ejmg.2014.01.006.)

278. Muriello M, Kim AY, Sondergaard Schatz K, Beck N, Gunay-Aygun M, Hoover-Fong JE: **Growth hormone deficiency, aortic dilation, and neurocognitive issues in Feingold syndrome 2**. *American journal of medical genetics Part A* 2019, **179**(3):410-416 [doi.org/10.1002/ajmg.a.61037.](https://doi.org/10.1002/ajmg.a.61037.)

279. Fares-Taie L, Gerber S, Tawara A, Ramirez-Miranda A, Douet JY, Verdin H, Guilloux A, Zenteno JC, Kondo H, Moisset H *et al*: **Submicroscopic deletions at 13q32.1 cause congenital microcoria**. *American journal of human genetics* 2015, **96**(4):631-639 [doi.org/10.1016/j.ajhg.2015.01.014.](https://doi.org/10.1016/j.ajhg.2015.01.014.)

280. Al-Owaid A, Alarfaj M, Al-Qahtani A, Al-Arfaj K: **Congenital microcoria in a Saudi family**. *Ophthalmic genetics* 2019, **40**(6):578-580 [doi.org/10.1080/13816810.2019.1692360.](https://doi.org/10.1080/13816810.2019.1692360.)

281. Pozza E, Verdin H, Deconinck H, Dheedene A, Menten B, De Baere E, Balikova I: **Microcoria due to first duplication of 13q32.1 including the GPR180 gene and maternal mosaicism**. *European journal of medical genetics* 2020, **63**(5):103918 [doi.org/10.1016/j.ejmg.2020.103918.](https://doi.org/10.1016/j.ejmg.2020.103918.)

282. Angée C, Nedelec B, Erjavec E, Rozet J-M, Fares Taie L: **Congenital Microcoria: Clinical Features and Molecular Genetics**. *Genes* 2021, **12**(5):624 [doi.org/10.3390/genes12050624.](https://doi.org/10.3390/genes12050624.)

283. Kirchhoff M, Bisgaard AM, Stoeva R, Dimitrov B, Gillessen-Kaesbach G, Fryns JP, Rose H, Grozdanova L, Ivanov I, Keymolen K *et al*: **Phenotype and 244k array-CGH characterization of chromosome 13q deletions: an update of the phenotypic map of 13q21.1-qter**. *American journal of medical genetics Part A* 2009, **149a**(5):894-905 [doi.org/10.1002/ajmg.a.32814.](https://doi.org/10.1002/ajmg.a.32814.)

284. Sagi-Dain L, Goldberg Y, Peleg A, Sukenik-Halevy R, Sofrin-Drucker E, Appelman Z, Josefsberg BYS, Ben-Shachar S, Vinkler C, Basel-Salmon L *et al*: **The rare 13q33–q34 microdeletions: eight new patients and review of the literature**. *Human genetics* 2019, **138**(10):1145-1153 [doi.org/10.1007/s00439-019-02048-y.](https://doi.org/10.1007/s00439-019-02048-y.)

285. Shimojima K, Komoike Y, Tohyama J, Takahashi S, Páez MT, Nakagawa E, Goto Y, Ohno K, Ohtsu M, Oguni H *et al*: **TULIP1 (RALGAPA1) haploinsufficiency with brain development delay**. *Genomics* 2009, **94**(6):414-422 [doi.org/10.1016/j.ygeno.2009.08.015.](https://doi.org/10.1016/j.ygeno.2009.08.015.)

286. Torgyekes E, Shanske AL, Anyane-Yeboa K, Nahum O, Pirzadeh S, Blumfield E, Jobanputra V, Warburton D, Levy B: **The proximal chromosome 14q microdeletion syndrome: delineation of the phenotype using high resolution SNP oligonucleotide microarray analysis (SOMA) and review of the literature**. *American journal of medical genetics Part A* 2011, **155a**(8):1884-1896 [doi.org/10.1002/ajmg.a.34090.](https://doi.org/10.1002/ajmg.a.34090.)

287. Fonseca DJ, Prada CF, Siza LM, Angel D, Gomez YM, Restrepo CM, Douben H, Rivadeneira F, de Klein A, Laissue P: **A de novo 14q12q13.3 interstitial deletion in a patient affected by a severe neurodevelopmental disorder of unknown origin**. *American journal of medical genetics Part A* 2012, **158a**(3):689-693 [doi.org/10.1002/ajmg.a.35215.](https://doi.org/10.1002/ajmg.a.35215.)

288. Santen GW, Sun Y, Gijsbers AC, Carré A, Holvoet M, Haeringen A, Lesnik Oberstein SA, Tomoda A, Mabe H, Polak M *et al*: **Further delineation of the phenotype of chromosome 14q13 deletions: (positional) involvement of FOXG1 appears the main determinant of phenotype severity, with no evidence for a holoprosencephaly locus**. *Journal of medical genetics* 2012, **49**(6):366-372 [doi.org/10.1136/jmedgenet-2011-100721.](https://doi.org/10.1136/jmedgenet-2011-100721.)

289. Gentile M, De Mattia D, Pansini A, Schettini F, Buonadonna AL, Capozza M, Ficarella R, Laforgia N: **14q13 distal microdeletion encompassing NKX2-1 and PAX9: Patient report and refinement of the associated phenotype**. *American Journal of Medical Genetics Part A* 2016, **170**(7):1884-1888 [doi.org/doi.org/10.1002/ajmg.a.37691.](https://doi.org/https://doi.org/10.1002/ajmg.a.37691.)

290. Brisset S, Slamova Z, Dusatkova P, Briand-Suleau A, Milcent K, Metay C, Simandlova M, Sumnik Z, Tosca L, Goossens M *et al*: **Anophthalmia, hearing loss, abnormal pituitary development and response to growth hormone therapy in three children with microdeletions of 14q22q23**. *Molecular cytogenetics* 2014, **7**(1):17 [doi.org/10.1186/1755-8166-7-17.](https://doi.org/10.1186/1755-8166-7-17.)

291. Martínez-Fernández ML, Bermejo-Sánchez E, Fernández B, MacDonald A, Fernández-Toral J, Martínez-Frías ML: **Haploinsufficiency of BMP4 gene may be the underlying cause of Frías syndrome**. *American journal of medical genetics Part A* 2014, **164a**(2):338-345 [doi.org/10.1002/ajmg.a.36224.](https://doi.org/10.1002/ajmg.a.36224.)

292. Blackburn PR, Zepeda-Mendoza CJ, Kruisselbrink TM, Schimmenti LA, García-Miñaur S, Palomares M, Nevado J, Mori MA, Le Meur G, Klee EW *et al*: **Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases**. *European Journal of Human Genetics* 2019, **27**(9):1379-1388 [doi.org/10.1038/s41431-019-0423-4.](https://doi.org/10.1038/s41431-019-0423-4.)

293. Kagami M, Nishimura G, Okuyama T, Hayashidani M, Takeuchi T, Tanaka S, Ishino F, Kurosawa K, Ogata T: **Segmental and full paternal isodisomy for chromosome 14 in three patients: narrowing the critical region and implication for the clinical features**. *American journal of medical genetics Part A* 2005, **138a**(2):127-132 [doi.org/10.1002/ajmg.a.30941.](https://doi.org/10.1002/ajmg.a.30941.)

294. Béna F, Gimelli S, Migliavacca E, Brun-Druc N, Buiting K, Antonarakis SE, Sharp AJ: **A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats**. *Hum Mol Genet* 2010, **19**(10):1967-1973 [doi.org/10.1093/hmg/ddq075.](https://doi.org/10.1093/hmg/ddq075.)

295. Zada A, Mundhofir FE, Pfundt R, Leijsten N, Nillesen W, Faradz SM, de Leeuw N: **A Rare, Recurrent, De Novo 14q32.2q32.31 Microdeletion of 1.1 Mb in a 20-Year-Old Female Patient with a Maternal UPD(14)-Like Phenotype and Intellectual Disability**. *Case Rep Genet* 2014, **2014**:530134 [doi.org/10.1155/2014/530134.](https://doi.org/10.1155/2014/530134.)

296. Severi G, Bernardini L, Briuglia S, Bigoni S, Buldrini B, Magini P, Dentici ML, Cordelli DM, Arrigo T, Franzoni E *et al*: **New patients with Temple syndrome caused by 14q32 deletion: Genotype-phenotype correlations and risk of thyroid cancer**. *American Journal of Medical Genetics Part A* 2016, **170**(1):162-169 [doi.org/doi.org/10.1002/ajmg.a.37346.](https://doi.org/https://doi.org/10.1002/ajmg.a.37346.)

297. Kagami M, Nagasaki K, Kosaki R, Horikawa R, Naiki Y, Saitoh S, Tajima T, Yorifuji T, Numakura C, Mizuno S *et al*: **Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients**. *Genetics in Medicine* 2017, **19**(12):1356-1366 [doi.org/10.1038/gim.2017.53.](https://doi.org/10.1038/gim.2017.53.)

298. Prasasya R, Grotheer KV, Siracusa LD, Bartolomei MS: **Temple syndrome and Kagami-Ogata syndrome: clinical presentations, genotypes, models and mechanisms**. *Hum Mol Genet* 2020, **29**(R1):R107-r116 [doi.org/10.1093/hmg/ddaa133.](https://doi.org/10.1093/hmg/ddaa133.)

299. Christian SL, Robinson WP, Huang B, Mutirangura A, Line MR, Nakao M, Surti U, Chakravarti A, Ledbetter DH: **Molecular characterization of two proximal deletion breakpoint regions in both Prader-Willi and Angelman syndrome patients**. *American journal of human genetics* 1995, **57**(1):40-48

300. Tan WH, Bacino CA, Skinner SA, Anselm I, Barbieri-Welge R, Bauer-Carlin A, Beaudet AL, Bichell TJ, Gentile JK, Glaze DG *et al*: **Angelman syndrome: Mutations influence features in early childhood**. *American journal of medical genetics Part A* 2011, **155a**(1):81-90 [doi.org/10.1002/ajmg.a.33775.](https://doi.org/10.1002/ajmg.a.33775.)

301. Kim SJ, Miller JL, Kuipers PJ, German JR, Beaudet AL, Sahoo T, Driscoll DJ: **Unique and atypical deletions in Prader-Willi syndrome reveal distinct phenotypes**. *Eur J Hum Genet* 2012, **20**(3):283-290 [doi.org/10.1038/ejhg.2011.187.](https://doi.org/10.1038/ejhg.2011.187.)

302. Cox DM, Butler MG: **The 15q11.2 BP1-BP2 microdeletion syndrome: a review**. *Int J Mol Sci* 2015, **16**(2):4068-4082 [doi.org/10.3390/ijms16024068.](https://doi.org/10.3390/ijms16024068.)

303. Burnside RD, Pasion R, Mikhail FM, Carroll AJ, Robin NH, Youngs EL, Gadi IK, Keitges E, Jaswaney VL, Papenhausen PR *et al*: **Microdeletion/microduplication of proximal 15q11.2 between BP1 and BP2: a susceptibility region for neurological dysfunction including developmental and language delay**. *Human genetics* 2011, **130**(4):517-528 [doi.org/10.1007/s00439-011-0970-4.](https://doi.org/10.1007/s00439-011-0970-4.)

304. von der Lippe C, Rustad C, Heimdal K, Rødningen OK: **15q11.2 microdeletion - seven new patients with delayed development and/or behavioural problems**. *European journal of medical genetics* 2011, **54**(3):357-360 [doi.org/10.1016/j.ejmg.2010.12.008.](https://doi.org/10.1016/j.ejmg.2010.12.008.)

305. Sahoo T, Theisen A, Rosenfeld JA, Lamb AN, Ravnan JB, Schultz RA, Torchia BS, Neill N, Casci I, Bejjani BA *et al*: **Copy number variants of schizophrenia susceptibility loci are associated with a spectrum of speech and developmental delays and behavior problems**. *Genet Med* 2011, **13**(10):868-880 [doi.org/10.1097/GIM.0b013e3182217a06.](https://doi.org/10.1097/GIM.0b013e3182217a06.)

306. Sharp AJ, Mefford HC, Li K, Baker C, Skinner C, Stevenson RE, Schroer RJ, Novara F, De Gregori M, Ciccone R *et al*: **A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures**. *Nature genetics* 2008, **40**(3):322-328 [doi.org/10.1038/ng.93.](https://doi.org/10.1038/ng.93.)

307. van Bon BW, Mefford HC, Menten B, Koolen DA, Sharp AJ, Nillesen WM, Innis JW, de Ravel TJ, Mercer CL, Fichera M *et al*: **Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome**. *Journal of medical genetics* 2009, **46**(8):511-523 [doi.org/10.1136/jmg.2008.063412.](https://doi.org/10.1136/jmg.2008.063412.)

308. Rosenfeld JA, Stephens LE, Coppinger J, Ballif BC, Hoo JJ, French BN, Banks VC, Smith WE, Manchester D, Tsai AC-H *et al*: **Deletions flanked by breakpoints 3 and 4 on 15q13 may contribute to abnormal phenotypes**. *Eur J Hum Genet* 2011, **19**(5):547-554 [doi.org/10.1038/ejhg.2010.237.](https://doi.org/10.1038/ejhg.2010.237.)

309. Miller DT, Shen Y, Weiss LA, Korn J, Anselm I, Bridgemohan C, Cox GF, Dickinson H, Gentile J, Harris DJ *et al*: **Microdeletion/duplication at 15q13.2q13.3 among individuals with features of autism and other neuropsychiatric disorders**. *Journal of medical genetics* 2009, **46**(4):242-248 [doi.org/10.1136/jmg.2008.059907.](https://doi.org/10.1136/jmg.2008.059907.)

310. Lowther C, Costain G, Stavropoulos DJ, Melvin R, Silversides CK, Andrade DM, So J, Faghfoury H, Lionel AC, Marshall CR *et al*: **Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature**. *Genet Med* 2015, **17**(2):149-157 [doi.org/10.1038/gim.2014.83.](https://doi.org/10.1038/gim.2014.83.)

311. Uddin M, Unda BK, Kwan V, Holzapfel NT, White SH, Chalil L, Woodbury-Smith M, Ho KS, Harward E, Murtaza N *et al*: **OTUD7A Regulates Neurodevelopmental Phenotypes in the 15q13.3 Microdeletion Syndrome**. *American journal of human genetics* 2018, **102**(2):278-295 [doi.org/10.1016/j.ajhg.2018.01.006.](https://doi.org/10.1016/j.ajhg.2018.01.006.)

312. Hoppman-Chaney N, Wain K, Seger PR, Superneau DW, Hodge JC: **Identification of single gene deletions at 15q13.3: further evidence that CHRNA7 causes the 15q13.3 microdeletion syndrome phenotype**. *Clin Genet* 2013, **83**(4):345-351 [doi.org/10.1111/j.1399-0004.2012.01925.x.](https://doi.org/10.1111/j.1399-0004.2012.01925.x.)

313. Johansson S, Berland S, Gradek GA, Bongers E, de Leeuw N, Pfundt R, Fannemel M, Rødningen O, Brendehaug A, Haukanes BI *et al*: **Haploinsufficiency of MEIS2 is associated with orofacial clefting and learning disability**. *American journal of medical genetics Part A* 2014, **164a**(7):1622-1626 [doi.org/10.1002/ajmg.a.36498.](https://doi.org/10.1002/ajmg.a.36498.)

314. Chen C-P, Chen C-Y, Chern S-R, Wu P-S, Chen Y-N, Chen S-W, Chen L-F, Yang C-W, Wang W: **Prenatal diagnosis and molecular cytogenetic characterization of a de novo 4.858-Mb microdeletion in 15q14 associated with ACTC1 and MEIS2 haploinsufficiency and tetralogy of Fallot**. *Taiwanese Journal of Obstetrics and Gynecology* 2016, **55**(2):270-274 [doi.org/doi.org/10.1016/j.tjog.2016.02.013.](https://doi.org/https://doi.org/10.1016/j.tjog.2016.02.013.)

315. Gambin T, Yuan B, Bi W, Liu P, Rosenfeld JA, Coban-Akdemir Z, Pursley AN, Nagamani SCS, Marom R, Golla S *et al*: **Identification of novel candidate disease genes from de novo exonic copy number variants**. *Genome Medicine* 2017, **9**(1):83 [doi.org/10.1186/s13073-017-0472-7.](https://doi.org/10.1186/s13073-017-0472-7.)

316. Shimojima K, Ondo Y, Okamoto N, Yamamoto T: **A 15q14 microdeletion involving MEIS2 identified in a patient with autism spectrum disorder**. *Human Genome Variation* 2017, **4**(1):17029 [doi.org/10.1038/hgv.2017.29.](https://doi.org/10.1038/hgv.2017.29.)

317. Zhang Y, Malekpour M, Al-Madani N, Kahrizi K, Zanganeh M, Lohr NJ, Mohseni M, Mojahedi F, Daneshi A, Najmabadi H *et al*: **Sensorineural deafness and male infertility: a contiguous gene deletion syndrome**. *Journal of medical genetics* 2007, **44**(4):233-240 [doi.org/10.1136/jmg.2006.045765.](https://doi.org/10.1136/jmg.2006.045765.)

318. Knijnenburg J, Oberstein SAJL, Frei K, Lucas T, Gijsbers ACJ, Ruivenkamp CAL, Tanke HJ, Szuhai K: **A homozygous deletion of a normal variation locus in a patient with hearing loss from non-consanguineous parents**. *Journal of medical genetics* 2009, **46**(6):412 [doi.org/10.1136/jmg.2008.063685.](https://doi.org/10.1136/jmg.2008.063685.)

319. Vona B, Hofrichter MA, Neuner C, Schröder J, Gehrig A, Hennermann JB, Kraus F, Shehata-Dieler W, Klopocki E, Nanda I *et al*: **DFNB16 is a frequent cause of congenital hearing impairment: implementation of STRC mutation analysis in routine diagnostics**. *Clin Genet* 2015, **87**(1):49-55 [doi.org/10.1111/cge.12332.](https://doi.org/10.1111/cge.12332.)

320. Mátyás G, Alonso S, Patrignani A, Marti M, Arnold E, Magyar I, Henggeler C, Carrel T, Steinmann B, Berger W: **Large genomic fibrillin-1 (FBN1) gene deletions provide evidence for true haploinsufficiency in Marfan syndrome**. *Human genetics* 2007, **122**(1):23-32 [doi.org/10.1007/s00439-007-0371-x.](https://doi.org/10.1007/s00439-007-0371-x.)

321. Furtado LV, Wooderchak-Donahue W, Rope AF, Yetman AT, Lewis T, Plant P, Bayrak-Toydemir P: **Characterization of large genomic deletions in the FBN1 gene using multiplex ligation-dependent probe amplification**. *BMC Medical Genetics* 2011, **12**(1):119 [doi.org/10.1186/1471-2350-12-119.](https://doi.org/10.1186/1471-2350-12-119.)

322. Hilhorst-Hofstee Y, Hamel BC, Verheij JB, Rijlaarsdam ME, Mancini GM, Cobben JM, Giroth C, Ruivenkamp CA, Hansson KB, Timmermans J *et al*: **The clinical spectrum of complete FBN1 allele deletions**. *Eur J Hum Genet* 2011, **19**(3):247-252 [doi.org/10.1038/ejhg.2010.174.](https://doi.org/10.1038/ejhg.2010.174.)

323. Dordoni C, Ciaccio C, Santoro G, Venturini M, Cavallari U, Ritelli M, Colombi M: **Marfan syndrome: Report of a complex phenotype due to a 15q21.1 contiguos gene deletion encompassing FBN1, and literature review**. *American Journal of Medical Genetics Part A* 2017, **173**(1):200-206 [doi.org/doi.org/10.1002/ajmg.a.37975.](https://doi.org/https://doi.org/10.1002/ajmg.a.37975.)

324. Magoulas PL, El-Hattab AW: **Chromosome 15q24 microdeletion syndrome**. *Orphanet J Rare Dis* 2012, **7**:2-2 [doi.org/10.1186/1750-1172-7-2.](https://doi.org/10.1186/1750-1172-7-2.)

325. Huynh MT, Lambert AS, Tosca L, Petit F, Philippe C, Parisot F, Benoît V, Linglart A, Brisset S, Tran CT *et al*: **15q24.1 BP4-BP1 microdeletion unmasking paternally inherited functional polymorphisms combined with distal 15q24.2q24.3 duplication in a patient with epilepsy, psychomotor delay, overweight, ventricular arrhythmia**. *European journal of medical genetics* 2018, **61**(8):459-464 [doi.org/10.1016/j.ejmg.2018.03.005.](https://doi.org/10.1016/j.ejmg.2018.03.005.)

326. Liu Y, Zhang Y, Zarrei M, Dong R, Yang X, Zhao D, Scherer SW, Gai Z: **Refining critical regions in 15q24 microdeletion syndrome pertaining to autism**. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* 2020, **183**(4):217-226 [doi.org/doi.org/10.1002/ajmg.b.32778.](https://doi.org/https://doi.org/10.1002/ajmg.b.32778.)

327. El-Hattab AW, Smolarek TA, Walker ME, Schorry EK, Immken LL, Patel G, Abbott M-A, Lanpher BC, Ou Z, Kang S-HL *et al*: **Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping**. *Human genetics* 2009, **126**(4):589-602 [doi.org/10.1007/s00439-009-0706-x.](https://doi.org/10.1007/s00439-009-0706-x.)

328. El-Hattab AW, Zhang F, Maxim R, Christensen KM, Ward JC, Hines-Dowell S, Scaglia F, Lupski JR, Cheung SW: **Deletion and duplication of 15q24: Molecular mechanisms and potential modification by additional copy number variants**. *Genetics in Medicine* 2010, **12**(9):573-586 [doi.org/10.1097/GIM.0b013e3181eb9b4a.](https://doi.org/10.1097/GIM.0b013e3181eb9b4a.)

329. Mefford HC, Rosenfeld JA, Shur N, Slavotinek AM, Cox VA, Hennekam RC, Firth HV, Willatt L, Wheeler P, Morrow EM *et al*: **Further clinical and molecular delineation of the 15q24 microdeletion syndrome**. *Journal of medical genetics* 2012, **49**(2):110 [doi.org/10.1136/jmedgenet-2011-100499.](https://doi.org/10.1136/jmedgenet-2011-100499.)

330. Witteveen JS, Willemsen MH, Dombroski TC, van Bakel NH, Nillesen WM, van Hulten JA, Jansen EJ, Verkaik D, Veenstra-Knol HE, van Ravenswaaij-Arts CM *et al*: **Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity**. *Nature genetics* 2016, **48**(8):877-887 [doi.org/10.1038/ng.3619.](https://doi.org/10.1038/ng.3619.)

331. Wat MJ, Enciso VB, Wiszniewski W, Resnick T, Bader P, Roeder ER, Freedenberg D, Brown C, Stankiewicz P, Cheung SW *et al*: **Recurrent microdeletions of 15q25.2 are associated with increased risk of congenital diaphragmatic hernia, cognitive deficits and possibly Diamond--Blackfan anaemia**. *Journal of medical genetics* 2010, **47**(11):777-781 [doi.org/10.1136/jmg.2009.075903.](https://doi.org/10.1136/jmg.2009.075903.)

332. Palumbo O, Palumbo P, Palladino T, Stallone R, Miroballo M, Piemontese MR, Zelante L, Carella M: **An emerging phenotype of interstitial 15q25.2 microdeletions: clinical report and review**. *American journal of medical genetics Part A* 2012, **158a**(12):3182-3189 [doi.org/10.1002/ajmg.a.35631.](https://doi.org/10.1002/ajmg.a.35631.)

333. Doelken SC, Seeger K, Hundsdoerfer P, Weber-Ferro W, Klopocki E, Graul-Neumann L: **Proximal and distal 15q25.2 microdeletions-genotype-phenotype delineation of two neurodevelopmental susceptibility loci**. *American journal of medical genetics Part A* 2013, **161a**(1):218-224 [doi.org/10.1002/ajmg.a.35695.](https://doi.org/10.1002/ajmg.a.35695.)

334. Tönnies H, Schulze I, Hennies H-C, Neumann LM, Keitzer R, Neitzel H: **De novo terminal deletion of chromosome 15q26.1 characterised by comparative genomic hybridisation and FISH with locus specific probes**. *Journal of medical genetics* 2001, **38**(9):617 [doi.org/10.1136/jmg.38.9.617.](https://doi.org/10.1136/jmg.38.9.617.)

335. Tatton-Brown K, Pilz DT, Örstavik KH, Patton M, Barber JCK, Collinson MN, Maloney VK, Huang S, Crolla JA, Marks K *et al*: **15q overgrowth syndrome: A newly recognized phenotype associated with overgrowth, learning difficulties, characteristic facial appearance, renal anomalies and increased dosage of distal chromosome 15q**. *American Journal of Medical Genetics Part A* 2009, **149A**(2):147-154 [doi.org/doi.org/10.1002/ajmg.a.32534.](https://doi.org/https://doi.org/10.1002/ajmg.a.32534.)

336. Chen C-P, Lin Y-H, Au H-K, Su Y-N, Hsu C-Y, Liu Y-P, Wu P-C, Chern S-R, Chen Y-T, Chen L-F *et al*: **Chromosome 15q overgrowth syndrome: Prenatal diagnosis, molecular cytogenetic characterization, and perinatal findings in a fetus with dup(15)(q26.2q26.3)**. *Taiwanese Journal of Obstetrics and Gynecology* 2011, **50**(3):359-365 [doi.org/doi.org/10.1016/j.tjog.2011.07.004.](https://doi.org/https://doi.org/10.1016/j.tjog.2011.07.004.)

337. Levy B, Tegay D, Papenhausen P, Tepperberg J, Nahum O, Tsuchida T, Pletcher BA, Ala-Kokko L, Baker S, Frederick B *et al*: **Tetrasomy 15q26: a distinct syndrome or Shprintzen-Goldberg syndrome phenocopy?** *Genet Med* 2012, **14**(9):811-818 [doi.org/10.1038/gim.2012.54.](https://doi.org/10.1038/gim.2012.54.)

338. Poot M, Verrijn Stuart AA, van Daalen E, van Iperen A, van Binsbergen E, Hochstenbach R: **Variable behavioural phenotypes of patients with monosomies of 15q26 and a review of 16 cases**. *European journal of medical genetics* 2013, **56**(7):346-350 [doi.org/10.1016/j.ejmg.2013.04.001.](https://doi.org/10.1016/j.ejmg.2013.04.001.)

339. Leffler M, Puusepp S, Žilina O, Zhu Y, Kuuse K, Bain N, Burgess T, Õunap K, Field M: **Two familial microduplications of 15q26.3 causing overgrowth and variable intellectual disability with normal copy number of IGF1R**. *European journal of medical genetics* 2016, **59**(4):257-262 [doi.org/10.1016/j.ejmg.2015.12.002.](https://doi.org/10.1016/j.ejmg.2015.12.002.)

340. Cannarella R, Mattina T, Condorelli RA, Mongioì LM, Pandini G, La Vignera S, Calogero AE: **Chromosome 15 structural abnormalities: effect on IGF1R gene expression and function**. *Endocr Connect* 2017, **6**(7):528-539 [doi.org/10.1530/ec-17-0158.](https://doi.org/10.1530/ec-17-0158.)

341. Oyazato Y, Iijima K, Emi M, Sekine T, Kamei K, Takanashi J, Nakao H, Namai Y, Nozu K, Matsuo M: **Molecular analysis of TSC2/PKD1 contiguous gene deletion syndrome**. *The Kobe journal of medical sciences* 2011, **57**(1):E1-10

342. Osumi K, Suga K, Ono A, Goji A, Mori T, Kinoshita Y, Sugano M, Toda Y, Urushihara M, Nakagawa R *et al*: **Molecular diagnosis of an infant with TSC2/PKD1 contiguous gene syndrome**. *Human Genome Variation* 2020, **7**(1):21 [doi.org/10.1038/s41439-020-0108-0.](https://doi.org/10.1038/s41439-020-0108-0.)

343. Hao YH, Fountain MD, Jr., Fon Tacer K, Xia F, Bi W, Kang SH, Patel A, Rosenfeld JA, Le Caignec C, Isidor B *et al*: **USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder**. *Molecular cell* 2015, **59**(6):956-969 [doi.org/10.1016/j.molcel.2015.07.033.](https://doi.org/10.1016/j.molcel.2015.07.033.)

344. Fountain MD, Oleson DS, Rech ME, Segebrecht L, Hunter JV, McCarthy JM, Lupo PJ, Holtgrewe M, Moran R, Rosenfeld JA *et al*: **Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies**. *Genet Med* 2019, **21**(8):1797-1807 [doi.org/10.1038/s41436-019-0433-1.](https://doi.org/10.1038/s41436-019-0433-1.)

345. Ullmann R, Turner G, Kirchhoff M, Chen W, Tonge B, Rosenberg C, Field M, Vianna-Morgante AM, Christie L, Krepischi-Santos AC *et al*: **Array CGH identifies reciprocal 16p13.1 duplications and deletions that predispose to autism and/or mental retardation**. *Human mutation* 2007, **28**(7):674-682 [doi.org/doi.org/10.1002/humu.20546.](https://doi.org/https://doi.org/10.1002/humu.20546.)

346. Hannes FD, Sharp AJ, Mefford HC, de Ravel T, Ruivenkamp CA, Breuning MH, Fryns JP, Devriendt K, Van Buggenhout G, Vogels A *et al*: **Recurrent reciprocal deletions and duplications of 16p13.11: the deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant**. *Journal of medical genetics* 2009, **46**(4):223-232 [doi.org/10.1136/jmg.2007.055202.](https://doi.org/10.1136/jmg.2007.055202.)

347. de Kovel CG, Trucks H, Helbig I, Mefford HC, Baker C, Leu C, Kluck C, Muhle H, von Spiczak S, Ostertag P *et al*: **Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies**. *Brain* 2010, **133**(Pt 1):23-32 [doi.org/10.1093/brain/awp262.](https://doi.org/10.1093/brain/awp262.)

348. Ingason A, Rujescu D, Cichon S, Sigurdsson E, Sigmundsson T, Pietiläinen OP, Buizer-Voskamp JE, Strengman E, Francks C, Muglia P *et al*: **Copy number variations of chromosome 16p13.1 region associated with schizophrenia**. *Mol Psychiatry* 2011, **16**(1):17-25 [doi.org/10.1038/mp.2009.101.](https://doi.org/10.1038/mp.2009.101.)

349. Ballif BC, Hornor SA, Jenkins E, Madan-Khetarpal S, Surti U, Jackson KE, Asamoah A, Brock PL, Gowans GC, Conway RL *et al*: **Discovery of a previously unrecognized microdeletion syndrome of 16p11.2-p12.2**. *Nature genetics* 2007, **39**(9):1071-1073 [doi.org/10.1038/ng2107.](https://doi.org/10.1038/ng2107.)

350. Battaglia A, Novelli A, Bernardini L, Igliozzi R, Parrini B: **Further characterization of the new microdeletion syndrome of 16p11.2-p12.2**. *American journal of medical genetics Part A* 2009, **149a**(6):1200-1204 [doi.org/10.1002/ajmg.a.32847.](https://doi.org/10.1002/ajmg.a.32847.)

351. Hempel M, Rivera Brugués N, Wagenstaller J, Lederer G, Weitensteiner A, Seidel H, Meitinger T, Strom TM: **Microdeletion syndrome 16p11.2-p12.2: clinical and molecular characterization**. *American journal of medical genetics Part A* 2009, **149a**(10):2106-2112 [doi.org/10.1002/ajmg.a.33042.](https://doi.org/10.1002/ajmg.a.33042.)

352. Tabet A-C, Pilorge M, Delorme R, Amsellem F, Pinard J-M, Leboyer M, Verloes A, Benzacken B, Betancur C: **Autism multiplex family with 16p11.2p12.2 microduplication syndrome in monozygotic twins and distal 16p11.2 deletion in their brother**. *European Journal of Human Genetics* 2012, **20**(5):540-546 [doi.org/10.1038/ejhg.2011.244.](https://doi.org/10.1038/ejhg.2011.244.)

353. Barber JC, Hall V, Maloney VK, Huang S, Roberts AM, Brady AF, Foulds N, Bewes B, Volleth M, Liehr T *et al*: **16p11.2-p12.2 duplication syndrome; a genomic condition differentiated from euchromatic variation of 16p11.2**. *Eur J Hum Genet* 2013, **21**(2):182-189 [doi.org/10.1038/ejhg.2012.144.](https://doi.org/10.1038/ejhg.2012.144.)

354. Ghebranious N, Giampietro PF, Wesbrook FP, Rezkalla SH: **A novel microdeletion at 16p11.2 harbors candidate genes for aortic valve development, seizure disorder, and mild mental retardation**. *American journal of medical genetics Part A* 2007, **143a**(13):1462-1471 [doi.org/10.1002/ajmg.a.31837.](https://doi.org/10.1002/ajmg.a.31837.)

355. Weiss LA, Shen Y, Korn JM, Arking DE, Miller DT, Fossdal R, Saemundsen E, Stefansson H, Ferreira MA, Green T *et al*: **Association between microdeletion and microduplication at 16p11.2 and autism**. *The New England journal of medicine* 2008, **358**(7):667-675 [doi.org/10.1056/NEJMoa075974.](https://doi.org/10.1056/NEJMoa075974.)

356. Bachmann-Gagescu R, Mefford HC, Cowan C, Glew GM, Hing AV, Wallace S, Bader PI, Hamati A, Reitnauer PJ, Smith R *et al*: **Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity**. *Genet Med* 2010, **12**(10):641-647 [doi.org/10.1097/GIM.0b013e3181ef4286.](https://doi.org/10.1097/GIM.0b013e3181ef4286.)

357. Bochukova EG, Huang N, Keogh J, Henning E, Purmann C, Blaszczyk K, Saeed S, Hamilton-Shield J, Clayton-Smith J, O'Rahilly S *et al*: **Large, rare chromosomal deletions associated with severe early-onset obesity**. *Nature* 2010, **463**(7281):666-670 [doi.org/10.1038/nature08689.](https://doi.org/10.1038/nature08689.)

358. Rosenfeld JA, Coppinger J, Bejjani BA, Girirajan S, Eichler EE, Shaffer LG, Ballif BC: **Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications**. *J Neurodev Disord* 2010, **2**(1):26-38 [doi.org/10.1007/s11689-009-9037-4.](https://doi.org/10.1007/s11689-009-9037-4.)

359. Sampson MG, Coughlin CR, 2nd, Kaplan P, Conlin LK, Meyers KE, Zackai EH, Spinner NB, Copelovitch L: **Evidence for a recurrent microdeletion at chromosome 16p11.2 associated with congenital anomalies of the kidney and urinary tract (CAKUT) and Hirschsprung disease**. *American journal of medical genetics Part A* 2010, **152a**(10):2618-2622 [doi.org/10.1002/ajmg.a.33628.](https://doi.org/10.1002/ajmg.a.33628.)

360. Rosenfeld JA, Coe BP, Eichler EE, Cuckle H, Shaffer LG: **Estimates of penetrance for recurrent pathogenic copy-number variations**. *Genet Med* 2013, **15**(6):478-481 [doi.org/10.1038/gim.2012.164.](https://doi.org/10.1038/gim.2012.164.)

361. Abdullah S, Helal M, Dupuis L, Stavropoulos DJ, Louro P, Ramos L, Mendoza-Londono R: **16q22.1 microdeletion and anticipatory guidance**. *American Journal of Medical Genetics Part A* 2019, **179**(7):1287-1292 [doi.org/doi.org/10.1002/ajmg.a.61155.](https://doi.org/https://doi.org/10.1002/ajmg.a.61155.)

362. Stankiewicz P, Sen P, Bhatt SS, Storer M, Xia Z, Bejjani BA, Ou Z, Wiszniewska J, Driscoll DJ, Maisenbacher MK *et al*: **Genomic and genic deletions of the FOX gene cluster on 16q24.1 and inactivating mutations of FOXF1 cause alveolar capillary dysplasia and other malformations**. *American journal of human genetics* 2009, **84**(6):780-791 [doi.org/10.1016/j.ajhg.2009.05.005.](https://doi.org/10.1016/j.ajhg.2009.05.005.)

363. Alsina Casanova M, Monteagudo-Sánchez A, Rodiguez Guerineau L, Court F, Gazquez Serrano I, Martorell L, Rovira Zurriaga C, Moore GE, Ishida M, Castañon M *et al*: **Maternal mutations of FOXF1 cause alveolar capillary dysplasia despite not being imprinted**. *Human mutation* 2017, **38**(6):615-620 [doi.org/doi.org/10.1002/humu.23213.](https://doi.org/https://doi.org/10.1002/humu.23213.)

364. Szafranski P, Kośmider E, Liu Q, Karolak JA, Currie L, Parkash S, Kahler SG, Roeder E, Littlejohn RO, DeNapoli TS *et al*: **LINE- and Alu-containing genomic instability hotspot at 16q24.1 associated with recurrent and nonrecurrent CNV deletions causative for ACDMPV**. *Human mutation* 2018, **39**(12):1916-1925 [doi.org/doi.org/10.1002/humu.23608.](https://doi.org/https://doi.org/10.1002/humu.23608.)

365. Szafranski P, Liu Q, Karolak JA, Song X, de Leeuw N, Faas B, Gerychova R, Janku P, Jezova M, Valaskova I *et al*: **Association of rare non-coding SNVs in the lung-specific FOXF1 enhancer with a mitigation of the lethal ACDMPV phenotype**. *Human genetics* 2019, **138**(11):1301-1311 [doi.org/10.1007/s00439-019-02073-x.](https://doi.org/10.1007/s00439-019-02073-x.)

366. Bi W, Sapir T, Shchelochkov OA, Zhang F, Withers MA, Hunter JV, Levy T, Shinder V, Peiffer DA, Gunderson KL *et al*: **Increased LIS1 expression affects human and mouse brain development**. *Nature genetics* 2009, **41**(2):168-177 [doi.org/10.1038/ng.302.](https://doi.org/10.1038/ng.302.)

367. Nagamani SC, Zhang F, Shchelochkov OA, Bi W, Ou Z, Scaglia F, Probst FJ, Shinawi M, Eng C, Hunter JV *et al*: **Microdeletions including YWHAE in the Miller-Dieker syndrome region on chromosome 17p13.3 result in facial dysmorphisms, growth restriction, and cognitive impairment**. *Journal of medical genetics* 2009, **46**(12):825-833 [doi.org/10.1136/jmg.2009.067637.](https://doi.org/10.1136/jmg.2009.067637.)

368. Roos L, Jønch AE, Kjaergaard S, Taudorf K, Simonsen H, Hamborg-Petersen B, Brøndum-Nielsen K, Kirchhoff M: **A new microduplication syndrome encompassing the region of the Miller–Dieker (17p13 deletion) syndrome**. *Journal of medical genetics* 2009, **46**(10):703 [doi.org/10.1136/jmg.2008.065094.](https://doi.org/10.1136/jmg.2008.065094.)

369. Bruno DL, Anderlid BM, Lindstrand A, van Ravenswaaij-Arts C, Ganesamoorthy D, Lundin J, Martin CL, Douglas J, Nowak C, Adam MP *et al*: **Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes**. *Journal of medical genetics* 2010, **47**(5):299-311 [doi.org/10.1136/jmg.2009.069906.](https://doi.org/10.1136/jmg.2009.069906.)

370. Curry CJ, Rosenfeld JA, Grant E, Gripp KW, Anderson C, Aylsworth AS, Saad TB, Chizhikov VV, Dybose G, Fagerberg C *et al*: **The duplication 17p13.3 phenotype: analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes**. *American journal of medical genetics Part A* 2013, **161A**(8):1833-1852 [doi.org/10.1002/ajmg.a.35996.](https://doi.org/10.1002/ajmg.a.35996.)

371. Barros Fontes MI, Dos Santos AP, Rossi Torres F, Lopes-Cendes I, Cendes F, Appenzeller S, Kawasaki de Araujo T, Lopes Monlleó I, Gil-da-Silva-Lopes VL: **17p13.3 Microdeletion: Insights on Genotype-Phenotype Correlation**. *Mol Syndromol* 2017, **8**(1):36-41 [doi.org/10.1159/000452753.](https://doi.org/10.1159/000452753.)

372. Emrick LT, Rosenfeld JA, Lalani SR, Jain M, Desai NK, Larson A, Kripps K, Vanderver A, Taft RJ, Bluske K *et al*: **Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum**. *Genet Med* 2019, **21**(7):1652-1656 [doi.org/10.1038/s41436-018-0358-0.](https://doi.org/10.1038/s41436-018-0358-0.)

373. Armour CM, Bulman DE, Jarinova O, Rogers RC, Clarkson KB, DuPont BR, Dwivedi A, Bartel FO, McDonell L, Schwartz CE *et al*: **17p13.3 microduplications are associated with split-hand/foot malformation and long-bone deficiency (SHFLD)**. *Eur J Hum Genet* 2011, **19**(11):1144-1151 [doi.org/10.1038/ejhg.2011.97.](https://doi.org/10.1038/ejhg.2011.97.)

374. Klopocki E, Lohan S, Doelken SC, Stricker S, Ockeloen CW, Soares Thiele de Aguiar R, Lezirovitz K, Mingroni Netto RC, Jamsheer A, Shah H *et al*: **Duplications of BHLHA9 are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion**. *Journal of medical genetics* 2012, **49**(2):119-125 [doi.org/10.1136/jmedgenet-2011-100409.](https://doi.org/10.1136/jmedgenet-2011-100409.)

375. Luk HM, Wong VC, Lo IF, Chan KY, Lau ET, Kan AS, Tang MH, Tang WF, She WM, Chu YW *et al*: **A prenatal case of split-hand malformation associated with 17p13.3 triplication - a dilemma in genetic counseling**. *European journal of medical genetics* 2014, **57**(2-3):81-84 [doi.org/10.1016/j.ejmg.2013.12.005.](https://doi.org/10.1016/j.ejmg.2013.12.005.)

376. Petit F, Jourdain AS, Andrieux J, Baujat G, Baumann C, Beneteau C, David A, Faivre L, Gaillard D, Gilbert-Dussardier B *et al*: **Split hand/foot malformation with long-bone deficiency and BHLHA9 duplication: report of 13 new families**. *Clinical Genetics* 2014, **85**(5):464-469 [doi.org/doi.org/10.1111/cge.12219.](https://doi.org/https://doi.org/10.1111/cge.12219.)

377. Shlien A, Baskin B, Achatz MI, Stavropoulos DJ, Nichols KE, Hudgins L, Morel CF, Adam MP, Zhukova N, Rotin L *et al*: **A common molecular mechanism underlies two phenotypically distinct 17p13.1 microdeletion syndromes**. *American journal of human genetics* 2010, **87**(5):631-642 [doi.org/10.1016/j.ajhg.2010.10.007.](https://doi.org/10.1016/j.ajhg.2010.10.007.)

378. Carvalho CM, Vasanth S, Shinawi M, Russell C, Ramocki MB, Brown CW, Graakjaer J, Skytte AB, Vianna-Morgante AM, Krepischi AC *et al*: **Dosage changes of a segment at 17p13.1 lead to intellectual disability and microcephaly as a result of complex genetic interaction of multiple genes**. *American journal of human genetics* 2014, **95**(5):565-578 [doi.org/10.1016/j.ajhg.2014.10.006.](https://doi.org/10.1016/j.ajhg.2014.10.006.)

379. Giordano L, Palestra F, Giuffrida MG, Molinaro A, Iodice A, Bernardini L, La Boria P, Accorsi P, Novelli A: **17p13.1 microdeletion: genetic and clinical findings in a new patient with epilepsy and comparison with literature**. *American journal of medical genetics Part A* 2014, **164a**(1):225-230 [doi.org/10.1002/ajmg.a.36225.](https://doi.org/10.1002/ajmg.a.36225.)

380. Maini I, Ivanovski I, Iodice A, Rosato S, Pollazzon M, Mussini M, Belligni EF, Coutton C, Marinelli M, Barbieri V *et al*: **Endocrinological Abnormalities Are a Main Feature of 17p13.1 Microduplication Syndrome: A New Case and Literature Review**. *Mol Syndromol* 2016, **7**(6):337-343 [doi.org/10.1159/000450718.](https://doi.org/10.1159/000450718.)

381. Leka-Emiri S, Petrou V, Manolakos E, Thomaidis L, Fotinou A, Vlachopapadopoulou E, Michalacos S: **17p13.1 Microduplication Syndrome in a Child, Familial Short Stature, and Growth Hormone Deficiency: A Case Report and Review of the Literature**. *Molecular Syndromology* 2018, **9**(6):300-305 [doi.org/10.1159/000494681.](https://doi.org/10.1159/000494681.)

382. Yuan B, Harel T, Gu S, Liu P, Burglen L, Chantot-Bastaraud S, Gelowani V, Beck CR, Carvalho CM, Cheung SW *et al*: **Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome**. *American journal of human genetics* 2015, **97**(5):691-707 [doi.org/10.1016/j.ajhg.2015.10.003.](https://doi.org/10.1016/j.ajhg.2015.10.003.)

383. Yuan B, Neira J, Gu S, Harel T, Liu P, Briceño I, Elsea SH, Gómez A, Potocki L, Lupski JR: **Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smith–Magenis syndrome with evident peripheral neuropathy**. *Human genetics* 2016, **135**(10):1161-1174 [doi.org/10.1007/s00439-016-1703-5.](https://doi.org/10.1007/s00439-016-1703-5.)

384. Fernández-Hernández L, Navarro-Cobos MJ, Alcántara-Ortigoza MA, Ramos-Ángeles SE, Molina-Álvarez B, Díaz-Cuéllar S, Asch-Daich B, González-del Angel A: **Report of a patient with a de novo non-recurrent duplication of 17p11.2p12 and Yq11 deletion**. *Molecular cytogenetics* 2019, **12**(1):35 [doi.org/10.1186/s13039-019-0438-0.](https://doi.org/10.1186/s13039-019-0438-0.)

385. Beck CR, Carvalho CMB, Akdemir ZC, Sedlazeck FJ, Song X, Meng Q, Hu J, Doddapaneni H, Chong Z, Chen ES *et al*: **Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2**. *Cell* 2019, **176**(6):1310-1324.e1310 [doi.org/doi.org/10.1016/j.cell.2019.01.045.](https://doi.org/https://doi.org/10.1016/j.cell.2019.01.045.)

386. Zhang F, Seeman P, Liu P, Weterman MAJ, Gonzaga-Jauregui C, Towne CF, Batish SD, De Vriendt E, De Jonghe P, Rautenstrauss B *et al*: **Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability**. *The American Journal of Human Genetics* 2010, **86**(6):892-903 [doi.org/doi.org/10.1016/j.ajhg.2010.05.001.](https://doi.org/https://doi.org/10.1016/j.ajhg.2010.05.001.)

387. Shaw CJ, Withers MA, Lupski JR: **Uncommon Deletions of the Smith-Magenis Syndrome Region Can Be Recurrent When Alternate Low-Copy Repeats Act as Homologous Recombination Substrates**. *The American Journal of Human Genetics* 2004, **75**(1):75-81 [doi.org/doi.org/10.1086/422016.](https://doi.org/https://doi.org/10.1086/422016.)

388. Potocki L, Bi W, Treadwell-Deering D, Carvalho CM, Eifert A, Friedman EM, Glaze D, Krull K, Lee JA, Lewis RA *et al*: **Characterization of Potocki-Lupski syndrome (dup(17)(p11.2p11.2)) and delineation of a dosage-sensitive critical interval that can convey an autism phenotype**. *American journal of human genetics* 2007, **80**(4):633-649 [doi.org/10.1086/512864.](https://doi.org/10.1086/512864.)

389. Zhang F, Potocki L, Sampson JB, Liu P, Sanchez-Valle A, Robbins-Furman P, Navarro AD, Wheeler PG, Spence JE, Brasington CK *et al*: **Identification of uncommon recurrent Potocki-Lupski syndrome-associated duplications and the distribution of rearrangement types and mechanisms in PTLS**. *American journal of human genetics* 2010, **86**(3):462-470 [doi.org/10.1016/j.ajhg.2010.02.001.](https://doi.org/10.1016/j.ajhg.2010.02.001.)

390. Franciskovich R, Soler-Alfonso C, Neira-Fresneda J, Lupski JR, McCann-Crosby B, Potocki L: **Short stature and growth hormone deficiency in a subset of patients with Potocki-Lupski syndrome: Expanding the phenotype of PTLS**. *American journal of medical genetics Part A* 2020, **182**(9):2077-2084 [doi.org/10.1002/ajmg.a.61741.](https://doi.org/10.1002/ajmg.a.61741.)

391. Kehrer-Sawatzki H, Mautner V-F, Cooper DN: **Emerging genotype-phenotype relationships in patients with large NF1 deletions**. *Human genetics* 2017, **136**(4):349-376 [doi.org/10.1007/s00439-017-1766-y.](https://doi.org/10.1007/s00439-017-1766-y.)

392. Moles KJ, Gowans GC, Gedela S, Beversdorf D, Yu A, Seaver LH, Schultz RA, Rosenfeld JA, Torchia BS, Shaffer LG: **NF1 microduplications: identification of seven nonrelated individuals provides further characterization of the phenotype**. *Genetics in Medicine* 2012, **14**(5):508-514 [doi.org/10.1038/gim.2011.46.](https://doi.org/10.1038/gim.2011.46.)

393. Serra G, Antona V, Corsello G, Zara F, Piro E, Falsaperla R: **NF1 microdeletion syndrome: case report of two new patients**. *Ital J Pediatr* 2019, **45**(1):138-138 [doi.org/10.1186/s13052-019-0718-7.](https://doi.org/10.1186/s13052-019-0718-7.)

394. Alvarado DM, Aferol H, McCall K, Huang JB, Techy M, Buchan J, Cady J, Gonzales PR, Dobbs MB, Gurnett CA: **Familial isolated clubfoot is associated with recurrent chromosome 17q23.1q23.2 microduplications containing TBX4**. *American journal of human genetics* 2010, **87**(1):154-160 [doi.org/10.1016/j.ajhg.2010.06.010.](https://doi.org/10.1016/j.ajhg.2010.06.010.)

395. Ballif BC, Theisen A, Rosenfeld JA, Traylor RN, Gastier-Foster J, Thrush DL, Astbury C, Bartholomew D, McBride KL, Pyatt RE *et al*: **Identification of a recurrent microdeletion at 17q23.1q23.2 flanked by segmental duplications associated with heart defects and limb abnormalities**. *American journal of human genetics* 2010, **86**(3):454-461 [doi.org/10.1016/j.ajhg.2010.01.038.](https://doi.org/10.1016/j.ajhg.2010.01.038.)

396. Schönewolf-Greulich B, Ronan A, Ravn K, Baekgaard P, Lodahl M, Nielsen K, Rendtorff ND, Tranebjaerg L, Brøndum-Nielsen K, Tümer Z: **Two new cases with microdeletion of 17q23.2 suggest presence of a candidate gene for sensorineural hearing loss within this region**. *American journal of medical genetics Part A* 2011, **155a**(12):2964-2969 [doi.org/10.1002/ajmg.a.34302.](https://doi.org/10.1002/ajmg.a.34302.)

397. Peterson JF, Ghaloul-Gonzalez L, Madan-Khetarpal S, Hartman J, Surti U, Rajkovic A, Yatsenko SA: **Familial microduplication of 17q23.1–q23.2 involving TBX4 is associated with congenital clubfoot and reduced penetrance in females**. *American journal of medical genetics Part A* 2014, **164a**(2):364-369 [doi.org/10.1002/ajmg.a.36238.](https://doi.org/10.1002/ajmg.a.36238.)

398. Sun M, Li N, Dong W, Chen Z, Liu Q, Xu Y, He G, Shi Y, Li X, Hao J *et al*: **Copy-number mutations on chromosome 17q24.2-q24.3 in congenital generalized hypertrichosis terminalis with or without gingival hyperplasia**. *American journal of human genetics* 2009, **84**(6):807-813 [doi.org/10.1016/j.ajhg.2009.04.018.](https://doi.org/10.1016/j.ajhg.2009.04.018.)

399. DeStefano GM, Kurban M, Anyane-Yeboa K, Dall'Armi C, Di Paolo G, Feenstra H, Silverberg N, Rohena L, López-Cepeda LD, Jobanputra V *et al*: **Mutations in the cholesterol transporter gene ABCA5 are associated with excessive hair overgrowth**. *PLoS Genet* 2014, **10**(5):e1004333 [doi.org/10.1371/journal.pgen.1004333.](https://doi.org/10.1371/journal.pgen.1004333.)

400. Afifi HH, Fukai R, Miyake N, Gamal El Din AA, Eid MM, Eid OM, Thomas MM, El-Badry TH, Tosson AM, Abdel-Salam GM *et al*: **De Novo 17q24.2-q24.3 microdeletion presenting with generalized hypertrichosis terminalis, gingival fibromatous hyperplasia, and distinctive facial features**. *American journal of medical genetics Part A* 2015, **167a**(10):2418-2424 [doi.org/10.1002/ajmg.a.37185.](https://doi.org/10.1002/ajmg.a.37185.)

401. Hayashi R, Yoshida K, Abe R, Niizeki H, Shimomura Y: **First Japanese case of congenital generalized hypertrichosis with a copy number variation on chromosome 17q24**. *Journal of dermatological science* 2017, **85**(1):63-65 [doi.org/10.1016/j.jdermsci.2016.10.010.](https://doi.org/10.1016/j.jdermsci.2016.10.010.)

402. Benko S, Fantes JA, Amiel J, Kleinjan D-J, Thomas S, Ramsay J, Jamshidi N, Essafi A, Heaney S, Gordon CT *et al*: **Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence**. *Nature genetics* 2009, **41**(3):359-364 [doi.org/10.1038/ng.329.](https://doi.org/10.1038/ng.329.)

403. Kurth I, Klopocki E, Stricker S, van Oosterwijk J, Vanek S, Altmann J, Santos HG, van Harssel JJT, de Ravel T, Wilkie AOM *et al*: **Duplications of noncoding elements 5′ of SOX9 are associated with brachydactyly-anonychia**. *Nature genetics* 2009, **41**(8):862-863 [doi.org/10.1038/ng0809-862.](https://doi.org/10.1038/ng0809-862.)

404. Benko S, Gordon CT, Mallet D, Sreenivasan R, Thauvin-Robinet C, Brendehaug A, Thomas S, Bruland O, David M, Nicolino M *et al*: **Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development**. *Journal of medical genetics* 2011, **48**(12):825-830 [doi.org/10.1136/jmedgenet-2011-100255.](https://doi.org/10.1136/jmedgenet-2011-100255.)

405. Vetro A, Ciccone R, Giorda R, Patricelli MG, Della Mina E, Forlino A, Zuffardi O: **XX males SRY negative: a confirmed cause of infertility**. *Journal of medical genetics* 2011, **48**(10):710-712 [doi.org/10.1136/jmedgenet-2011-100036.](https://doi.org/10.1136/jmedgenet-2011-100036.)

406. Kim GJ, Sock E, Buchberger A, Just W, Denzer F, Hoepffner W, German J, Cole T, Mann J, Seguin JH *et al*: **Copy number variation of two separate regulatory regions upstream of SOX9 causes isolated 46,XY or 46,XX disorder of sex development**. *Journal of medical genetics* 2015, **52**(4):240-247 [doi.org/10.1136/jmedgenet-2014-102864.](https://doi.org/10.1136/jmedgenet-2014-102864.)

407. Wester U, Bondeson ML, Edeby C, Annerén G: **Clinical and molecular characterization of individuals with 18p deletion: a genotype-phenotype correlation**. *American journal of medical genetics Part A* 2006, **140**(11):1164-1171 [doi.org/10.1002/ajmg.a.31260.](https://doi.org/10.1002/ajmg.a.31260.)

408. Turleau C: **Monosomy 18p**. *Orphanet J Rare Dis* 2008, **3**:4-4 [doi.org/10.1186/1750-1172-3-4.](https://doi.org/10.1186/1750-1172-3-4.)

409. Sebold C, Roeder E, Zimmerman M, Soileau B, Heard P, Carter E, Schatz M, White WA, Perry B, Reinker K *et al*: **Tetrasomy 18p: report of the molecular and clinical findings of 43 individuals**. *American journal of medical genetics Part A* 2010, **152a**(9):2164-2172 [doi.org/10.1002/ajmg.a.33597.](https://doi.org/10.1002/ajmg.a.33597.)

410. O'Donnell L, Soileau BT, Sebold C, Gelfond J, Hale DE, Cody JD: **Tetrasomy 18p: report of cognitive and behavioral characteristics**. *American journal of medical genetics Part A* 2015, **167**(7):1474-1482 [doi.org/10.1002/ajmg.a.37036.](https://doi.org/10.1002/ajmg.a.37036.)

411. Cody JD, Sebold C, Malik A, Heard P, Carter E, Crandall A, Soileau B, Semrud-Clikeman M, Cody CM, Hardies LJ *et al*: **Recurrent interstitial deletions of proximal 18q: a new syndrome involving expressive speech delay**. *American journal of medical genetics Part A* 2007, **143a**(11):1181-1190 [doi.org/10.1002/ajmg.a.31729.](https://doi.org/10.1002/ajmg.a.31729.)

412. Feenstra I, Vissers LE, Orsel M, van Kessel AG, Brunner HG, Veltman JA, van Ravenswaaij-Arts CM: **Genotype-phenotype mapping of chromosome 18q deletions by high-resolution array CGH: an update of the phenotypic map**. *American journal of medical genetics Part A* 2007, **143a**(16):1858-1867 [doi.org/10.1002/ajmg.a.31850.](https://doi.org/10.1002/ajmg.a.31850.)

413. Heard PL, Carter EM, Crandall AC, Sebold C, Hale DE, Cody JD: **High resolution genomic analysis of 18q- using oligo-microarray comparative genomic hybridization (aCGH)**. *American journal of medical genetics Part A* 2009, **149A**(7):1431-1437 [doi.org/10.1002/ajmg.a.32900.](https://doi.org/10.1002/ajmg.a.32900.)

414. Cody JD, Sebold C, Heard P, Carter E, Soileau B, Hasi-Zogaj M, Hill A, Rupert D, Perry B, O'Donnell L *et al*: **Consequences of chromsome18q deletions**. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* 2015, **169**(3):265-280 [doi.org/doi.org/10.1002/ajmg.c.31446.](https://doi.org/https://doi.org/10.1002/ajmg.c.31446.)

415. Rojnueangnit K, Charalsawadi C, Thammachote W, Pradabmuksiri A, Tim-Aroon T, Novelli A, Loddo S, Briuglia S, Concetta CM, Wattanasirichaigoon D *et al*: **Clinical delineation of 18q11-q12 microdeletion: Intellectual disability, speech and behavioral disorders, and conotruncal heart defects**. *Molecular genetics & genomic medicine* 2019, **7**(9):e896-e896 [doi.org/10.1002/mgg3.896.](https://doi.org/10.1002/mgg3.896.)

416. Hasi M, Soileau B, Sebold C, Hill A, Hale DE, O’Donnell L, Cody JD: **The role of the TCF4 gene in the phenotype of individuals with 18q segmental deletions**. *Human genetics* 2011, **130**(6):777-787 [doi.org/10.1007/s00439-011-1020-y.](https://doi.org/10.1007/s00439-011-1020-y.)

417. Cody JD, Hasi M, Soileau B, Heard P, Carter E, Sebold C, O’Donnell L, Perry B, Stratton RF, Hale DE: **Establishing a reference group for distal 18q-: clinical description and molecular basis**. *Human genetics* 2014, **133**(2):199-209 [doi.org/10.1007/s00439-013-1364-6.](https://doi.org/10.1007/s00439-013-1364-6.)

418. Ishikawa A, Enomoto K, Tominaga M, Saito T, Nagai J, Furuya N, Ueno K, Ueda H, Masuno M, Kurosawa K: **Pure duplication of 19p13.3**. *American journal of medical genetics Part A* 2013, **161a**(9):2300-2304 [doi.org/10.1002/ajmg.a.36041.](https://doi.org/10.1002/ajmg.a.36041.)

419. Nevado J, Rosenfeld JA, Mena R, Palomares-Bralo M, Vallespín E, Ángeles Mori M, Tenorio JA, Gripp KW, Denenberg E, Del Campo M *et al*: **PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome**. *Eur J Hum Genet* 2015, **23**(12):1615-1626 [doi.org/10.1038/ejhg.2015.51.](https://doi.org/10.1038/ejhg.2015.51.)

420. Orellana C, Roselló M, Monfort S, Mayo S, Oltra S, Martínez F: **Pure duplication of 19p13.3 in three members of a family with intellectual disability and literature review. Definition of a new microduplication syndrome**. *American Journal of Medical Genetics Part A* 2015, **167**(7):1614-1620 [doi.org/doi.org/10.1002/ajmg.a.37046.](https://doi.org/https://doi.org/10.1002/ajmg.a.37046.)

421. Novikova I, Sen P, Manzardo A, Butler MG: **Duplication of 19p13.3 in 11-Year-Old Male Patient with Dysmorphic Features and Intellectual Disability: A Review**. *J Pediatr Genet* 2017, **6**(4):227-233 [doi.org/10.1055/s-0037-1603650.](https://doi.org/10.1055/s-0037-1603650.)

422. Tenorio J, Nevado J, González-Meneses A, Arias P, Dapía I, Venegas-Vega CA, Calvente M, Hernández A, Landera L, Ramos S *et al*: **Further definition of the proximal 19p13.3 microdeletion/microduplication syndrome and implication of PIAS4 as the major contributor**. *Clinical Genetics* 2020, **97**(3):467-476 [doi.org/doi.org/10.1111/cge.13689.](https://doi.org/https://doi.org/10.1111/cge.13689.)

423. Dolan M, Mendelsohn NJ, Pierpont ME, Schimmenti LA, Berry SA, Hirsch B: **A novel microdeletion/microduplication syndrome of 19p13.13**. *Genetics in Medicine* 2010, **12**(8):503-511 [doi.org/10.1097/GIM.0b013e3181e59291.](https://doi.org/10.1097/GIM.0b013e3181e59291.)

424. Malan V, Rajan D, Thomas S, Shaw AC, Louis Dit Picard H, Layet V, Till M, van Haeringen A, Mortier G, Nampoothiri S *et al*: **Distinct effects of allelic NFIX mutations on nonsense-mediated mRNA decay engender either a Sotos-like or a Marshall-Smith syndrome**. *American journal of human genetics* 2010, **87**(2):189-198 [doi.org/10.1016/j.ajhg.2010.07.001.](https://doi.org/10.1016/j.ajhg.2010.07.001.)

425. Shimojima K, Okamoto N, Tamasaki A, Sangu N, Shimada S, Yamamoto T: **An association of 19p13.2 microdeletions with Malan syndrome and Chiari malformation**. *American Journal of Medical Genetics Part A* 2015, **167**(4):724-730 [doi.org/doi.org/10.1002/ajmg.a.36959.](https://doi.org/https://doi.org/10.1002/ajmg.a.36959.)

426. Jezela-Stanek A, Kucharczyk M, Falana K, Jurkiewicz D, Mlynek M, Wicher D, Rydzanicz M, Kugaudo M, Cieslikowska A, Ciara E *et al*: **Malan syndrome (Sotos syndrome 2) in two patients with 19p13.2 deletion encompassing NFIX gene and novel NFIX sequence variant**. *Biomedical papers of the Medical Faculty of the University Palacky, Olomouc, Czechoslovakia* 2016, **160**(1):161-167 [doi.org/10.5507/bp.2016.006.](https://doi.org/10.5507/bp.2016.006.)

427. Jensen DR, Martin DM, Gebarski S, Sahoo T, Brundage EK, Chinault AC, Otto EA, Chaki M, Hildebrandt F, Cheung SW *et al*: **A novel chromosome 19p13.12 deletion in a child with multiple congenital anomalies**. *American journal of medical genetics Part A* 2009, **149A**(3):396-402 [doi.org/10.1002/ajmg.a.32691.](https://doi.org/10.1002/ajmg.a.32691.)

428. Bonaglia MC, Marelli S, Novara F, Commodaro S, Borgatti R, Minardo G, Memo L, Mangold E, Beri S, Zucca C *et al*: **Genotype-phenotype relationship in three cases with overlapping 19p13.12 microdeletions**. *Eur J Hum Genet* 2010, **18**(12):1302-1309 [doi.org/10.1038/ejhg.2010.115.](https://doi.org/10.1038/ejhg.2010.115.)

429. Gallant NM, Baldwin E, Salamon N, Dipple KM, Quintero-Rivera F: **Pontocerebellar hypoplasia in association with de novo 19p13.11p13.12 microdeletion**. *American journal of medical genetics Part A* 2011, **155a**(11):2871-2878 [doi.org/10.1002/ajmg.a.34286.](https://doi.org/10.1002/ajmg.a.34286.)

430. Van der Aa N, Vandeweyer G, Kooy RF: **A boy with mental retardation, obesity and hypertrichosis caused by a microdeletion of 19p13.12**. *European journal of medical genetics* 2010, **53**(5):291-293 [doi.org/10.1016/j.ejmg.2010.05.006.](https://doi.org/10.1016/j.ejmg.2010.05.006.)

431. de Souza LC, Sgardioli IC, Gil-da-Silva-Lopes VL, Vieira TP: **A recognizable phenotype related to 19p13.12 microdeletion**. *American Journal of Medical Genetics Part A* 2018, **176**(8):1753-1759 [doi.org/doi.org/10.1002/ajmg.a.38842.](https://doi.org/https://doi.org/10.1002/ajmg.a.38842.)

432. Chowdhury S, Bandholz AM, Parkash S, Dyack S, Rideout AL, Leppig KA, Thiese H, Wheeler PG, Tsang M, Ballif BC *et al*: **Phenotypic and molecular characterization of 19q12q13.1 deletions: a report of five patients**. *American journal of medical genetics Part A* 2014, **164a**(1):62-69 [doi.org/10.1002/ajmg.a.36201.](https://doi.org/10.1002/ajmg.a.36201.)

433. Caubit X, Gubellini P, Andrieux J, Roubertoux PL, Metwaly M, Jacq B, Fatmi A, Had-Aissouni L, Kwan KY, Salin P *et al*: **TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons**. *Nature genetics* 2016, **48**(11):1359-1369 [doi.org/10.1038/ng.3681.](https://doi.org/10.1038/ng.3681.)

434. Malan V, Raoul O, Firth HV, Royer G, Turleau C, Bernheim A, Willatt L, Munnich A, Vekemans M, Lyonnet S *et al*: **19q13.11 deletion syndrome: a novel clinically recognisable genetic condition identified by array comparative genomic hybridisation**. *Journal of medical genetics* 2009, **46**(9):635-640 [doi.org/10.1136/jmg.2008.062034.](https://doi.org/10.1136/jmg.2008.062034.)

435. Gana S, Veggiotti P, Sciacca G, Fedeli C, Bersano A, Micieli G, Maghnie M, Ciccone R, Rossi E, Plunkett K *et al*: **19q13.11 cryptic deletion: description of two new cases and indication for a role of WTIP haploinsufficiency in hypospadias**. *European Journal of Human Genetics* 2012, **20**(8):852-856 [doi.org/10.1038/ejhg.2012.19.](https://doi.org/10.1038/ejhg.2012.19.)

436. Abe KT, Rizzo IMPO, Coelho ALV, Sakai N, Jr., Carvalho DR, Speck-Martins CE: **19q13.11 microdeletion: Clinical features overlapping ectrodactyly ectodermal dysplasia-clefting syndrome phenotype**. *Clin Case Rep* 2018, **6**(7):1300-1307 [doi.org/10.1002/ccr3.1600.](https://doi.org/10.1002/ccr3.1600.)

437. Yuan H, Meng Z, Liu L, Deng X, Hu X, Liang L: **A de novo 1.6Mb microdeletion at 19q13.2 in a boy with Diamond-Blackfan anemia, global developmental delay and multiple congenital anomalies**. *Molecular cytogenetics* 2016, **9**:58 [doi.org/10.1186/s13039-016-0268-2.](https://doi.org/10.1186/s13039-016-0268-2.)

438. Kessi M, Xiong J, Wu L, Yang L, He F, Chen C, Pang N, Duan H, Zhang W, Arafat A *et al*: **Rare Copy Number Variations and Predictors in Children With Intellectual Disability and Epilepsy**. *Front Neurol* 2018, **9**:947-947 [doi.org/10.3389/fneur.2018.00947.](https://doi.org/10.3389/fneur.2018.00947.)

439. Calpena E, McGowan SJ, Blanco Kelly F, Boudry-Labis E, Dieux-Coeslier A, Harrison R, Johnson D, Lachlan K, Morton JEV, Stewart H *et al*: **Dissection of contiguous gene effects for deletions around ERF on chromosome 19**. *Human mutation* 2021, **n/a**(n/a) [doi.org/doi.org/10.1002/humu.24213.](https://doi.org/https://doi.org/10.1002/humu.24213.)

440. Singh R, Cohen ASA, Poulton C, Hjortshøj TD, Akahira-Azuma M, Mendiratta G, Khan WA, Azmanov DN, Woodward KJ, Kirchhoff M *et al*: **Deletion of ERF and CIC causes abnormal skull morphology and global developmental delay**. *Cold Spring Harbor molecular case studies* 2021, **7**(3) [doi.org/10.1101/mcs.a005991.](https://doi.org/10.1101/mcs.a005991.)

441. McGill AK, Pastore MT, Herman GE, Alliman S, Rosenfeld JA, Weaver DD: **A tale of two deletions: A report of two novel 20p13 → pter deletions**. *American Journal of Medical Genetics Part A* 2010, **152A**(4):1000-1007 [doi.org/doi.org/10.1002/ajmg.a.33339.](https://doi.org/https://doi.org/10.1002/ajmg.a.33339.)

442. Moutton S, Rooryck C, Toutain J, Cailley D, Bouron J, Villega F, Taupiac E, Lacombe D, Arveiler B, Goizet C: **Dysmorphic features in subtelomeric 20p13 deletion excluding JAG1: A recognizable microdeletion phenotype?** *European journal of medical genetics* 2012, **55**(2):151-155 [doi.org/doi.org/10.1016/j.ejmg.2011.12.009.](https://doi.org/https://doi.org/10.1016/j.ejmg.2011.12.009.)

443. An Y, Amr SS, Torres A, Weissman L, Raffalli P, Cox G, Sheng X, Lip V, Bi W, Patel A *et al*: **SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay**. *American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics* 2013, **162b**(8):832-840 [doi.org/10.1002/ajmg.b.32187.](https://doi.org/10.1002/ajmg.b.32187.)

444. Fang H-H, Liu S-Y, Wang Y-F, Chiang C-M, Liu C-C, Lin C-M: **Phenotypic features of a microdeletion in chromosome band 20p13: A case report and review of the literature**. *Molecular Genetics & Genomic Medicine* 2019, **7**(7):e00739 [doi.org/doi.org/10.1002/mgg3.739.](https://doi.org/https://doi.org/10.1002/mgg3.739.)

445. Lalani SR, Thakuria JV, Cox GF, Wang X, Bi W, Bray MS, Shaw C, Cheung SW, Chinault AC, Boggs BA *et al*: **20p12.3 microdeletion predisposes to Wolff-Parkinson-White syndrome with variable neurocognitive deficits**. *Journal of medical genetics* 2009, **46**(3):168-175 [doi.org/10.1136/jmg.2008.061002.](https://doi.org/10.1136/jmg.2008.061002.)

446. Sahoo T, Theisen A, Sanchez-Lara PA, Marble M, Schweitzer DN, Torchia BS, Lamb AN, Bejjani BA, Shaffer LG, Lacassie Y: **Microdeletion 20p12.3 involving BMP2 contributes to syndromic forms of cleft palate**. *American journal of medical genetics Part A* 2011, **155A**(7):1646-1653 [doi.org/10.1002/ajmg.a.34063.](https://doi.org/10.1002/ajmg.a.34063.)

447. Williams ES, Uhas KA, Bunke BP, Garber KB, Martin CL: **Cleft palate in a multigenerational family with a microdeletion of 20p12.3 involving BMP2**. *American Journal of Medical Genetics Part A* 2012, **158A**(10):2616-2620 [doi.org/doi.org/10.1002/ajmg.a.35594.](https://doi.org/https://doi.org/10.1002/ajmg.a.35594.)

448. Tan TY, Gonzaga-Jauregui C, Bhoj EJ, Strauss KA, Brigatti K, Puffenberger E, Li D, Xie L, Das N, Skubas I *et al*: **Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions**. *The American Journal of Human Genetics* 2017, **101**(6):985-994 [doi.org/doi.org/10.1016/j.ajhg.2017.10.006.](https://doi.org/https://doi.org/10.1016/j.ajhg.2017.10.006.)

449. Traylor RN, Bruno DL, Burgess T, Wildin R, Spencer A, Ganesamoorthy D, Amor DJ, Hunter M, Caplan M, Rosenfeld JA *et al*: **A genotype-first approach for the molecular and clinical characterization of uncommon de novo microdeletion of 20q13.33**. *PLoS One* 2010, **5**(8):e12462 [doi.org/10.1371/journal.pone.0012462.](https://doi.org/10.1371/journal.pone.0012462.)

450. Mefford HC, Cook J, Gospe Jr SM: **Epilepsy due to 20q13.33 subtelomere deletion masquerading as pyridoxine-dependent epilepsy**. *American Journal of Medical Genetics Part A* 2012, **158A**(12):3190-3195 [doi.org/doi.org/10.1002/ajmg.a.35633.](https://doi.org/https://doi.org/10.1002/ajmg.a.35633.)

451. Mosca-Boidron AL, Valduga M, Thauvin-Robinet C, Lagarde N, Marle N, Henry C, Pinoit JM, Huet F, Béri-Deixheimer M, Ragon C *et al*: **Additional evidence to support the role of the 20q13.33 region in susceptibility to autism**. *American journal of medical genetics Part A* 2013, **161a**(6):1505-1507 [doi.org/10.1002/ajmg.a.35878.](https://doi.org/10.1002/ajmg.a.35878.)

452. Oegema R, de Klein A, Verkerk AJ, Schot R, Dumee B, Douben H, Eussen B, Dubbel L, Poddighe PJ, van der Laar I *et al*: **Distinctive Phenotypic Abnormalities Associated with Submicroscopic 21q22 Deletion Including DYRK1A**. *Molecular syndromology* 2010, **1**(3):113-120 [doi.org/10.1159/000320113.](https://doi.org/10.1159/000320113.)

453. Valetto A, Orsini A, Bertini V, Toschi B, Bonuccelli A, Simi F, Sammartino I, Taddeucci G, Simi P, Saggese G: **Molecular cytogenetic characterization of an interstitial deletion of chromosome 21 (21q22.13q22.3) in a patient with dysmorphic features, intellectual disability and severe generalized epilepsy**. *European journal of medical genetics* 2012, **55**(5):362-366 [doi.org/10.1016/j.ejmg.2012.03.011.](https://doi.org/10.1016/j.ejmg.2012.03.011.)

454. Ji J, Lee H, Argiropoulos B, Dorrani N, Mann J, Martinez-Agosto JA, Gomez-Ospina N, Gallant N, Bernstein JA, Hudgins L *et al*: **DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies**. *European Journal of Human Genetics* 2015, **23**(11):1473-1481 [doi.org/10.1038/ejhg.2015.71.](https://doi.org/10.1038/ejhg.2015.71.)

455. McDermid HE, Morrow BE: **Genomic disorders on 22q11**. *American journal of human genetics* 2002, **70**(5):1077-1088 [doi.org/10.1086/340363.](https://doi.org/10.1086/340363.)

456. Kriek M, Szuhai K, Kant SG, White SJ, Dauwerse H, Fiegler H, Carter NP, Knijnenburg J, den Dunnen JT, Tanke HJ *et al*: **A complex rearrangement on chromosome 22 affecting both homologues; haplo-insufficiency of the Cat eye syndrome region may have no clinical relevance**. *Human genetics* 2006, **120**(1):77-84 [doi.org/10.1007/s00439-006-0185-2.](https://doi.org/10.1007/s00439-006-0185-2.)

457. Zhang J, Ma D, Wang Y, Cao L, Wu Y, Qiao F, Liu A, Li L, Lin Y, Liu G *et al*: **Analysis of chromosome 22q11 copy number variations by multiplex ligation-dependent probe amplification for prenatal diagnosis of congenital heart defect**. *Molecular cytogenetics* 2015, **8**:100 [doi.org/10.1186/s13039-015-0209-5.](https://doi.org/10.1186/s13039-015-0209-5.)

458. Gottlieb S, Driscoll DA, Punnett HH, Sellinger B, Emanuel BS, Budarf ML: **Characterization of 10p deletions suggests two nonoverlapping regions contribute to the DiGeorge syndrome phenotype**. *American journal of human genetics* 1998, **62**(2):495-498 [doi.org/10.1086/301718.](https://doi.org/10.1086/301718.)

459. Burnside RD: **22q11.21 Deletion Syndromes: A Review of Proximal, Central, and Distal Deletions and Their Associated Features**. *Cytogenetic and Genome Research* 2015, **146**(2):89-99 [doi.org/10.1159/000438708.](https://doi.org/10.1159/000438708.)

460. Morrow BE, McDonald-McGinn DM, Emanuel BS, Vermeesch JR, Scambler PJ: **Molecular genetics of 22q11.2 deletion syndrome**. *American journal of medical genetics Part A* 2018, **176**(10):2070-2081 [doi.org/10.1002/ajmg.a.40504.](https://doi.org/10.1002/ajmg.a.40504.)

461. Rump P, de Leeuw N, van Essen AJ, Verschuuren-Bemelmans CC, Veenstra-Knol HE, Swinkels MEM, Oostdijk W, Ruivenkamp C, Reardon W, de Munnik S *et al*: **Central 22q11.2 deletions**. *American Journal of Medical Genetics Part A* 2014, **164**(11):2707-2723 [doi.org/doi.org/10.1002/ajmg.a.36711.](https://doi.org/https://doi.org/10.1002/ajmg.a.36711.)

462. Woodward KJ, Stampalia J, Vanyai H, Rijhumal H, Potts K, Taylor F, Peverall J, Grumball T, Sivamoorthy S, Alinejad-Rokny H *et al*: **Atypical nested 22q11.2 duplications between LCR22B and LCR22D are associated with neurodevelopmental phenotypes including autism spectrum disorder with incomplete penetrance**. *Mol Genet Genomic Med* 2019, **7**(2):e00507 [doi.org/10.1002/mgg3.507.](https://doi.org/10.1002/mgg3.507.)

463. Rauch A, Zink S, Zweier C, Thiel CT, Koch A, Rauch R, Lascorz J, Hüffmeier U, Weyand M, Singer H *et al*: **Systematic assessment of atypical deletions reveals genotype-phenotype correlation in 22q11.2**. *Journal of medical genetics* 2005, **42**(11):871-876 [doi.org/10.1136/jmg.2004.030619.](https://doi.org/10.1136/jmg.2004.030619.)

464. Ben-Shachar S, Ou Z, Shaw CA, Belmont JW, Patel MS, Hummel M, Amato S, Tartaglia N, Berg J, Sutton VR *et al*: **22q11.2 distal deletion: a recurrent genomic disorder distinct from DiGeorge syndrome and velocardiofacial syndrome**. *American journal of human genetics* 2008, **82**(1):214-221 [doi.org/10.1016/j.ajhg.2007.09.014.](https://doi.org/10.1016/j.ajhg.2007.09.014.)

465. Tan TY, Collins A, James PA, McGillivray G, Stark Z, Gordon CT, Leventer RJ, Pope K, Forbes R, Crolla JA *et al*: **Phenotypic variability of distal 22q11.2 copy number abnormalities**. *American journal of medical genetics Part A* 2011, **155a**(7):1623-1633 [doi.org/10.1002/ajmg.a.34051.](https://doi.org/10.1002/ajmg.a.34051.)

466. Mikhail FM, Burnside RD, Rush B, Ibrahim J, Godshalk R, Rutledge SL, Robin NH, Descartes MD, Carroll AJ: **The recurrent distal 22q11.2 microdeletions are often de novo and do not represent a single clinical entity: a proposed categorization system**. *Genet Med* 2014, **16**(1):92-100 [doi.org/10.1038/gim.2013.79.](https://doi.org/10.1038/gim.2013.79.)

467. Dhar SU, del Gaudio D, German JR, Peters SU, Ou Z, Bader PI, Berg JS, Blazo M, Brown CW, Graham BH *et al*: **22q13.3 deletion syndrome: Clinical and molecular analysis using array CGH**. *American Journal of Medical Genetics Part A* 2010, **152A**(3):573-581 [doi.org/doi.org/10.1002/ajmg.a.33253.](https://doi.org/https://doi.org/10.1002/ajmg.a.33253.)

468. Phelan K, McDermid HE: **The 22q13.3 Deletion Syndrome (Phelan-McDermid Syndrome)**. *Molecular Syndromology* 2011, **2**(3-5):186-201 [doi.org/10.1159/000334260.](https://doi.org/10.1159/000334260.)

469. Han K, Holder JL, Jr., Schaaf CP, Lu H, Chen H, Kang H, Tang J, Wu Z, Hao S, Cheung SW *et al*: **SHANK3 overexpression causes manic-like behaviour with unique pharmacogenetic properties**. *Nature* 2013, **503**(7474):72-77 [doi.org/10.1038/nature12630.](https://doi.org/10.1038/nature12630.)

470. Disciglio V, Lo Rizzo C, Mencarelli MA, Mucciolo M, Marozza A, Di Marco C, Massarelli A, Canocchi V, Baldassarri M, Ndoni E *et al*: **Interstitial 22q13 deletions not involving SHANK3 gene: a new contiguous gene syndrome**. *American journal of medical genetics Part A* 2014, **164a**(7):1666-1676 [doi.org/10.1002/ajmg.a.36513.](https://doi.org/10.1002/ajmg.a.36513.)

471. Phelan K, Boccuto L, Rogers RC, Sarasua SM, McDermid HE: **Letter to the editor regarding Disciglio et al.: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome**. *American Journal of Medical Genetics Part A* 2015, **167**(7):1679-1680 [doi.org/doi.org/10.1002/ajmg.a.36788.](https://doi.org/https://doi.org/10.1002/ajmg.a.36788.)

472. Upadia J, Gonzales PR, Atkinson TP, Schroeder HW, Robin NH, Rudy NL, Mikhail FM: **A previously unrecognized 22q13.2 microdeletion syndrome that encompasses TCF20 and TNFRSF13C**. *American Journal of Medical Genetics Part A* 2018, **176**(12):2791-2797 [doi.org/doi.org/10.1002/ajmg.a.40492.](https://doi.org/https://doi.org/10.1002/ajmg.a.40492.)

473. Shapiro LJ, Yen P, Pomerantz D, Martin E, Rolewic L, Mohandas T: **Molecular studies of deletions at the human steroid sulfatase locus**. *Proceedings of the National Academy of Sciences of the United States of America* 1989, **86**(21):8477-8481 [doi.org/10.1073/pnas.86.21.8477.](https://doi.org/10.1073/pnas.86.21.8477.)

474. Alperin ES, Shapiro LJ: **Characterization of point mutations in patients with X-linked ichthyosis. Effects on the structure and function of the steroid sulfatase protein**. *The Journal of biological chemistry* 1997, **272**(33):20756-20763 [doi.org/10.1074/jbc.272.33.20756.](https://doi.org/10.1074/jbc.272.33.20756.)

475. Valdes-Flores M, Kofman-Alfaro SH, Vaca AL, Cuevas-Covarrubias SA: **Mutation report: a novel partial deletion of exons 2-10 of the STS gene in recessive X-linked ichthyosis**. *The Journal of investigative dermatology* 2000, **114**(3):591-593 [doi.org/10.1046/j.1523-1747.2000.00924.x.](https://doi.org/10.1046/j.1523-1747.2000.00924.x.)

476. Hand JL, Runke CK, Hodge JC: **The phenotype spectrum of X-linked ichthyosis identified by chromosomal microarray**. *J Am Acad Dermatol* 2015, **72**(4):617-627 [doi.org/10.1016/j.jaad.2014.12.020.](https://doi.org/10.1016/j.jaad.2014.12.020.)

477. Brcic L, Underwood JF, Kendall KM, Caseras X, Kirov G, Davies W: **Medical and neurobehavioural phenotypes in carriers of X-linked ichthyosis-associated genetic deletions in the UK Biobank**. *Journal of medical genetics* 2020, **57**(10):692-698 [doi.org/10.1136/jmedgenet-2019-106676.](https://doi.org/10.1136/jmedgenet-2019-106676.)

478. Gubb SJA, Brcic L, Underwood JFG, Kendall KM, Caseras X, Kirov G, Davies W: **Medical and neurobehavioural phenotypes in male and female carriers of Xp22.31 duplications in the UK Biobank**. *Hum Mol Genet* 2020, **29**(17):2872-2881 [doi.org/10.1093/hmg/ddaa174.](https://doi.org/10.1093/hmg/ddaa174.)

479. Hellerud C, Adamowicz M, Jurkiewicz D, Taybert J, Kubalska J, Ciara E, Popowska E, Ellis JR, Lindstedt S, Pronicka E: **Clinical heterogeneity and molecular findings in five Polish patients with glycerol kinase deficiency: investigation of two splice site mutations with computerized splice junction analysis and Xp21 gene-specific mRNA analysis**. *Mol Genet Metab* 2003, **79**(3):149-159 [doi.org/10.1016/s1096-7192](https://doi.org/10.1016/s1096-7192)(03)00094-5.

480. Stanczak CM, Chen Z, Zhang YH, Nelson SF, McCabe ER: **Deletion mapping in Xp21 for patients with complex glycerol kinase deficiency using SNP mapping arrays**. *Human mutation* 2007, **28**(3):235-242 [doi.org/10.1002/humu.20424.](https://doi.org/10.1002/humu.20424.)

481. Heide S, Afenjar A, Edery P, Sanlaville D, Keren B, Rouen A, Lavillaureix A, Hyon C, Doummar D, Siffroi JP *et al*: **Xp21 deletion in female patients with intellectual disability: Two new cases and a review of the literature**. *European journal of medical genetics* 2015, **58**(6-7):341-345 [doi.org/10.1016/j.ejmg.2015.04.003.](https://doi.org/10.1016/j.ejmg.2015.04.003.)

482. Korkut S, Baştuğ O, Raygada M, Hatipoğlu N, Kurtoğlu S, Kendirci M, Lyssikatos C, Stratakis CA: **Complex Glycerol Kinase Deficiency and Adrenocortical Insufficiency in Two Neonates**. *J Clin Res Pediatr Endocrinol* 2016, **8**(4):468-471 [doi.org/10.4274/jcrpe.2539.](https://doi.org/10.4274/jcrpe.2539.)

483. Montoya-Williams D, Mowitz M: **Cholestasis and Hepatic Iron Deposition in an Infant With Complex Glycerol Kinase Deficiency**. *Pediatrics* 2017, **140**(1):e20161479 [doi.org/10.1542/peds.2016-1479.](https://doi.org/10.1542/peds.2016-1479.)

484. Lugtenberg D, Yntema HG, Banning MJ, Oudakker AR, Firth HV, Willatt L, Raynaud M, Kleefstra T, Fryns JP, Ropers HH *et al*: **ZNF674: a new kruppel-associated box-containing zinc-finger gene involved in nonsyndromic X-linked mental retardation**. *American journal of human genetics* 2006, **78**(2):265-278 [doi.org/10.1086/500306.](https://doi.org/10.1086/500306.)

485. Zhang L, Wang T, Wright AF, Suri M, Schwartz CE, Stevenson RE, Valle D: **A microdeletion in Xp11.3 accounts for co-segregation of retinitis pigmentosa and mental retardation in a large kindred**. *American journal of medical genetics Part A* 2006, **140**(4):349-357 [doi.org/10.1002/ajmg.a.31080.](https://doi.org/10.1002/ajmg.a.31080.)

486. Delphin N, Hanein S, Taie LF, Zanlonghi X, Bonneau D, Moisan J-P, Boyle C, Nitschke P, Pruvost S, Bonnefont J-P *et al*: **Intellectual disability associated with retinal dystrophy in the Xp11.3 deletion syndrome: ZNF674 on trial. Guilty or innocent?** *Eur J Hum Genet* 2012, **20**(3):352-356 [doi.org/10.1038/ejhg.2011.217.](https://doi.org/10.1038/ejhg.2011.217.)

487. Giorda R, Bonaglia MC, Beri S, Fichera M, Novara F, Magini P, Urquhart J, Sharkey FH, Zucca C, Grasso R *et al*: **Complex segmental duplications mediate a recurrent dup(X)(p11.22-p11.23) associated with mental retardation, speech delay, and EEG anomalies in males and females**. *American journal of human genetics* 2009, **85**(3):394-400 [doi.org/10.1016/j.ajhg.2009.08.001.](https://doi.org/10.1016/j.ajhg.2009.08.001.)

488. Nizon M, Andrieux J, Rooryck C, de Blois MC, Bourel-Ponchel E, Bourgois B, Boute O, David A, Delobel B, Duban-Bedu B *et al*: **Phenotype-genotype correlations in 17 new patients with an Xp11.23p11.22 microduplication and review of the literature**. *American journal of medical genetics Part A* 2015, **167a**(1):111-122 [doi.org/10.1002/ajmg.a.36807.](https://doi.org/10.1002/ajmg.a.36807.)

489. Grams SE, Argiropoulos B, Lines M, Chakraborty P, McGowan-Jordan J, Geraghty MT, Tsang M, Eswara M, Tezcan K, Adams KL *et al*: **Genotype-phenotype characterization in 13 individuals with chromosome Xp11.22 duplications**. *American journal of medical genetics Part A* 2016, **170a**(4):967-977 [doi.org/10.1002/ajmg.a.37519.](https://doi.org/10.1002/ajmg.a.37519.)

490. Whibley AC, Plagnol V, Tarpey PS, Abidi F, Fullston T, Choma MK, Boucher CA, Shepherd L, Willatt L, Parkin G *et al*: **Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability**. *American journal of human genetics* 2010, **87**(2):173-188 [doi.org/10.1016/j.ajhg.2010.06.017.](https://doi.org/10.1016/j.ajhg.2010.06.017.)

491. Froyen G, Belet S, Martinez F, Santos-Rebouças CB, Declercq M, Verbeeck J, Donckers L, Berland S, Mayo S, Rosello M *et al*: **Copy-number gains of HUWE1 due to replication- and recombination-based rearrangements**. *American journal of human genetics* 2012, **91**(2):252-264 [doi.org/10.1016/j.ajhg.2012.06.010.](https://doi.org/10.1016/j.ajhg.2012.06.010.)

492. Song MH, Lee HK, Choi JY, Kim S, Bok J, Kim UK: **Clinical evaluation of DFN3 patients with deletions in the POU3F4 locus and detection of carrier female using MLPA**. *Clin Genet* 2010, **78**(6):524-532 [doi.org/10.1111/j.1399-0004.2010.01426.x.](https://doi.org/10.1111/j.1399-0004.2010.01426.x.)

493. Iossa S, Costa V, Corvino V, Auletta G, Barruffo L, Cappellani S, Ceglia C, Cennamo G, D'Adamo AP, D'Amico A *et al*: **Phenotypic and genetic characterization of a family carrying two Xq21.1-21.3 interstitial deletions associated with syndromic hearing loss**. *Molecular cytogenetics* 2015, **8**:18 [doi.org/10.1186/s13039-015-0120-0.](https://doi.org/10.1186/s13039-015-0120-0.)

494. Liang S, Jiang N, Li S, Jiang X, Yu D: **A maternally inherited 8.05 Mb Xq21 deletion associated with Choroideremia, deafness, and mental retardation syndrome in a male patient**. *Molecular cytogenetics* 2017, **10**:23-23 [doi.org/10.1186/s13039-017-0324-6.](https://doi.org/10.1186/s13039-017-0324-6.)

495. Meloni I, Vitelli F, Pucci L, Lowry RB, Tonlorenzi R, Rossi E, Ventura M, Rizzoni G, Kashtan CE, Pober B *et al*: **Alport syndrome and mental retardation: clinical and genetic dissection of the contiguous gene deletion syndrome in Xq22.3 (ATS-MR)**. *Journal of medical genetics* 2002, **39**(5):359-365 [doi.org/10.1136/jmg.39.5.359.](https://doi.org/10.1136/jmg.39.5.359.)

496. Uliana V, Marcocci E, Mucciolo M, Meloni I, Izzi C, Manno C, Bruttini M, Mari F, Scolari F, Renieri A *et al*: **Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2**. *Pediatric Nephrology* 2011, **26**(5):717-724 [doi.org/10.1007/s00467-010-1693-9.](https://doi.org/10.1007/s00467-010-1693-9.)

497. Nozu K, Minamikawa S, Yamada S, Oka M, Yanagita M, Morisada N, Fujinaga S, Nagano C, Gotoh Y, Takahashi E *et al*: **Characterization of contiguous gene deletions in COL4A6 and COL4A5 in Alport syndrome-diffuse leiomyomatosis**. *Journal of Human Genetics* 2017, **62**(7):733-735 [doi.org/10.1038/jhg.2017.28.](https://doi.org/10.1038/jhg.2017.28.)

498. Hoischen A, Landwehr C, Kabisch S, Ding X-Q, Trost D, Stropahl G, Wigger M, Radlwimmer B, Weber RG, Haffner D: **Array-CGH in unclear syndromic nephropathies identifies a microdeletion in Xq22.3-q23**. *Pediatric Nephrology* 2009, **24**(9):1673-1681 [doi.org/10.1007/s00467-009-1184-z.](https://doi.org/10.1007/s00467-009-1184-z.)

499. Rodriguez JD, Bhat SS, Meloni I, Ladd S, Leslie ND, Doyne EO, Renieri A, DuPont BR, Stevenson RE, Schwartz CE *et al*: **Intellectual disability, midface hypoplasia, facial hypotonia, and alport syndrome are associated with a deletion in Xq22.3**. *American Journal of Medical Genetics Part A* 2010, **152A**(3):713-717 [doi.org/doi.org/10.1002/ajmg.a.33208.](https://doi.org/https://doi.org/10.1002/ajmg.a.33208.)

500. Di Benedetto D, Musumeci SA, Avola E, Alberti A, Buono S, Scuderi C, Grillo L, Galesi O, Spalletta A, Giudice ML *et al*: **Definition of minimal duplicated region encompassing the XIAP and STAG2 genes in the Xq25 microduplication syndrome**. *American journal of medical genetics Part A* 2014, **164a**(8):1923-1930 [doi.org/10.1002/ajmg.a.36570.](https://doi.org/10.1002/ajmg.a.36570.)

501. Yingjun X, Wen T, Yujian L, Lingling X, Huimin H, Qun F, Junhong C: **Microduplication of chromosome Xq25 encompassing STAG2 gene in a boy with intellectual disability**. *European journal of medical genetics* 2015, **58**(2):116-121 [doi.org/doi.org/10.1016/j.ejmg.2014.10.002.](https://doi.org/https://doi.org/10.1016/j.ejmg.2014.10.002.)

502. Leroy C, Jacquemont ML, Doray B, Lamblin D, Cormier-Daire V, Philippe A, Nusbaum S, Patrat C, Steffann J, Colleaux L *et al*: **Xq25 duplication: the crucial role of the STAG2 gene in this novel human cohesinopathy**. *Clin Genet* 2016, **89**(1):68-73 [doi.org/10.1111/cge.12567.](https://doi.org/10.1111/cge.12567.)

503. Turchi G, Bernardo P, Consales A, Bilo L, Coppola A: **Xq25 microduplication syndrome: a further contribution to its definition. A case report and review of the literature**. *Clinical dysmorphology* 2020, **29**(2):90-96 [doi.org/10.1097/mcd.0000000000000303.](https://doi.org/10.1097/mcd.0000000000000303.)

504. Trivellin G, Daly AF, Faucz FR, Yuan B, Rostomyan L, Larco DO, Schernthaner-Reiter MH, Szarek E, Leal LF, Caberg JH *et al*: **Gigantism and acromegaly due to Xq26 microduplications and GPR101 mutation**. *The New England journal of medicine* 2014, **371**(25):2363-2374 [doi.org/10.1056/NEJMoa1408028.](https://doi.org/10.1056/NEJMoa1408028.)

505. Iacovazzo D, Caswell R, Bunce B, Jose S, Yuan B, Hernández-Ramírez LC, Kapur S, Caimari F, Evanson J, Ferraù F *et al*: **Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study**. *Acta neuropathologica communications* 2016, **4**(1):56 [doi.org/10.1186/s40478-016-0328-1.](https://doi.org/10.1186/s40478-016-0328-1.)

506. Naves LA, Daly AF, Dias LA, Yuan B, Zakir JC, Barra GB, Palmeira L, Villa C, Trivellin G, Júnior AJ *et al*: **Aggressive tumor growth and clinical evolution in a patient with X-linked acro-gigantism syndrome**. *Endocrine* 2016, **51**(2):236-244 [doi.org/10.1007/s12020-015-0804-6.](https://doi.org/10.1007/s12020-015-0804-6.)

507. Sutton E, Hughes J, White S, Sekido R, Tan J, Arboleda V, Rogers N, Knower K, Rowley L, Eyre H *et al*: **Identification of SOX3 as an XX male sex reversal gene in mice and humans**. *The Journal of clinical investigation* 2011, **121**(1):328-341 [doi.org/10.1172/jci42580.](https://doi.org/10.1172/jci42580.)

508. Moalem S, Babul-Hirji R, Stavropolous DJ, Wherrett D, Bägli DJ, Thomas P, Chitayat D: **XX male sex reversal with genital abnormalities associated with a de novo SOX3 gene duplication**. *American journal of medical genetics Part A* 2012, **158a**(7):1759-1764 [doi.org/10.1002/ajmg.a.35390.](https://doi.org/10.1002/ajmg.a.35390.)

509. Vetro A, Dehghani MR, Kraoua L, Giorda R, Beri S, Cardarelli L, Merico M, Manolakos E, Parada-Bustamante A, Castro A *et al*: **Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3**. *European Journal of Human Genetics* 2015, **23**(8):1025-1032 [doi.org/10.1038/ejhg.2014.237.](https://doi.org/10.1038/ejhg.2014.237.)

510. Tasic V, Mitrotti A, Riepe FG, Kulle AE, Laban N, Polenakovic M, Plaseska-Karanfilska D, Sanna-Cherchi S, Kostovski M, Gucev Z: **Duplication of The SOX3 Gene in an Sry-negative 46,XX Male with Associated Congenital Anomalies of Kidneys and the Urinary Tract: Case Report and Review of the Literature**. *Balkan J Med Genet* 2019, **22**(1):81-88 [doi.org/10.2478/bjmg-2019-0006.](https://doi.org/10.2478/bjmg-2019-0006.)

511. Bowl MR, Nesbit MA, Harding B, Levy E, Jefferson A, Volpi E, Rizzoti K, Lovell-Badge R, Schlessinger D, Whyte MP *et al*: **An interstitial deletion-insertion involving chromosomes 2p25.3 and Xq27.1, near SOX3, causes X-linked recessive hypoparathyroidism**. *The Journal of clinical investigation* 2005, **115**(10):2822-2831 [doi.org/10.1172/jci24156.](https://doi.org/10.1172/jci24156.)

512. Arya VB, Chawla G, Nambisan AKR, Muhi-Iddin N, Vamvakiti E, Ajzensztejn M, Hulse T, Ferreira Pinto C, Lahiri N, Bint S *et al*: **Xq27.1 Duplication Encompassing <b><i>SOX3</i></b>: Variable Phenotype and Smallest Duplication Associated with Hypopituitarism to Date – A Large Case Series of Unrelated Patients and a Literature Review**. *Hormone Research in Paediatrics* 2019, **92**(6):382-389 [doi.org/10.1159/000503784.](https://doi.org/10.1159/000503784.)

513. Rosolowsky ET, Stein R, Marks SD, Leonard N: **Marked phenotypic variable expression among brothers with duplication of Xq27.1 involving the SOX3 gene**. *Journal of pediatric endocrinology & metabolism : JPEM* 2020, **33**(3):443-447 [doi.org/10.1515/jpem-2015-0131.](https://doi.org/10.1515/jpem-2015-0131.)

514. Rio M, Malan V, Boissel S, Toutain A, Royer G, Gobin S, Morichon-Delvallez N, Turleau C, Bonnefont JP, Munnich A *et al*: **Familial interstitial Xq27.3q28 duplication encompassing the FMR1 gene but not the MECP2 gene causes a new syndromic mental retardation condition**. *Eur J Hum Genet* 2010, **18**(3):285-290 [doi.org/10.1038/ejhg.2009.159.](https://doi.org/10.1038/ejhg.2009.159.)

515. Nagamani SCS, Erez A, Probst FJ, Bader P, Evans P, Baker LA, Fang P, Bertin T, Hixson P, Stankiewicz P *et al*: **Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function**. *neurogenetics* 2012, **13**(4):333-339 [doi.org/10.1007/s10048-012-0340-y.](https://doi.org/10.1007/s10048-012-0340-y.)

516. Vengoechea J, Parikh AS, Zhang S, Tassone F: **De novo microduplication of the FMR1 gene in a patient with developmental delay, epilepsy and hyperactivity**. *European Journal of Human Genetics* 2012, **20**(11):1197-1200 [doi.org/10.1038/ejhg.2012.78.](https://doi.org/10.1038/ejhg.2012.78.)

517. Hickey SE, Walters-Sen L, Mosher TM, Pfau RB, Pyatt R, Snyder PJ, Sotos JF, Prior TW: **Duplication of the Xq27.3–q28 region, including the FMR1 gene, in an X-linked hypogonadism, gynecomastia, intellectual disability, short stature, and obesity syndrome**. *American Journal of Medical Genetics Part A* 2013, **161**(9):2294-2299 [doi.org/doi.org/10.1002/ajmg.a.36034.](https://doi.org/https://doi.org/10.1002/ajmg.a.36034.)

518. Amir RE, Van den Veyver IB, Wan M, Tran CQ, Francke U, Zoghbi HY: **Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2**. *Nature genetics* 1999, **23**(2):185-188 [doi.org/10.1038/13810.](https://doi.org/10.1038/13810.)

519. Van Esch H: **MECP2 Duplication Syndrome**. *Mol Syndromol* 2012, **2**(3-5):128-136 [doi.org/10.1159/000329580.](https://doi.org/10.1159/000329580.)

520. Vidal S, Pascual-Alonso A, Rabaza-Gairí M, Gerotina E, Brandi N, Pacheco P, Xiol C, Pineda M, Armstrong J: **Characterization of large deletions of the MECP2 gene in Rett syndrome patients by gene dosage analysis**. *Mol Genet Genomic Med* 2019, **7**(8):e793 [doi.org/10.1002/mgg3.793.](https://doi.org/10.1002/mgg3.793.)

521. Pascual-Alonso A, Blasco L, Vidal S, Gean E, Rubio P, O'Callaghan M, Martínez-Monseny AF, Castells AA, Xiol C, Català V *et al*: **Molecular characterization of Spanish patients with MECP2 duplication syndrome**. *Clin Genet* 2020, **97**(4):610-620 [doi.org/10.1111/cge.13718.](https://doi.org/10.1111/cge.13718.)

522. Froyen G, Van Esch H, Bauters M, Hollanders K, Frints SG, Vermeesch JR, Devriendt K, Fryns JP, Marynen P: **Detection of genomic copy number changes in patients with idiopathic mental retardation by high-resolution X-array-CGH: important role for increased gene dosage of XLMR genes**. *Human mutation* 2007, **28**(10):1034-1042 [doi.org/10.1002/humu.20564.](https://doi.org/10.1002/humu.20564.)

523. Madrigal I, Rodríguez-Revenga L, Armengol L, González E, Rodriguez B, Badenas C, Sánchez A, Martínez F, Guitart M, Fernández I *et al*: **X-chromosome tiling path array detection of copy number variants in patients with chromosome X-linked mental retardation**. *BMC genomics* 2007, **8**:443 [doi.org/10.1186/1471-2164-8-443.](https://doi.org/10.1186/1471-2164-8-443.)

524. Vandewalle J, Van Esch H, Govaerts K, Verbeeck J, Zweier C, Madrigal I, Mila M, Pijkels E, Fernandez I, Kohlhase J *et al*: **Dosage-dependent severity of the phenotype in patients with mental retardation due to a recurrent copy-number gain at Xq28 mediated by an unusual recombination**. *American journal of human genetics* 2009, **85**(6):809-822 [doi.org/10.1016/j.ajhg.2009.10.019.](https://doi.org/10.1016/j.ajhg.2009.10.019.)

525. Sinibaldi L, Parisi V, Lanciotti S, Fontana P, Kuechler A, Baujat G, Torres B, Koetting J, Splendiani A, Postorivo D *et al*: **Delineation of MidXq28-duplication syndrome distal to MECP2 and proximal to RAB39B genes**. *Clin Genet* 2019, **96**(3):246-253 [doi.org/10.1111/cge.13565.](https://doi.org/10.1111/cge.13565.)

526. El-Hattab AW, Fang P, Jin W, Hughes JR, Gibson JB, Patel GS, Grange DK, Manwaring LP, Patel A, Stankiewicz P *et al*: **Int22h-1/int22h-2-mediated Xq28 rearrangements: intellectual disability associated with duplications and in utero male lethality with deletions**. *Journal of medical genetics* 2011, **48**(12):840-850 [doi.org/10.1136/jmedgenet-2011-100125.](https://doi.org/10.1136/jmedgenet-2011-100125.)

527. Vanmarsenille L, Giannandrea M, Fieremans N, Verbeeck J, Belet S, Raynaud M, Vogels A, Männik K, Õunap K, Jacqueline V *et al*: **Increased dosage of RAB39B affects neuronal development and could explain the cognitive impairment in male patients with distal Xq28 copy number gains**. *Human mutation* 2014, **35**(3):377-383 [doi.org/10.1002/humu.22497.](https://doi.org/10.1002/humu.22497.)

528. El-Hattab AW, Schaaf CP, Fang P, Roeder E, Kimonis VE, Church JA, Patel A, Cheung SW: **Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review**. *BMC Med Genet* 2015, **16**:12 [doi.org/10.1186/s12881-015-0157-2.](https://doi.org/10.1186/s12881-015-0157-2.)

529. Hervé D, Touraine P, Verloes A, Miskinyte S, Krivosic V, Logeart D, Alili N, Laredo JD, Gaudric A, Houdart E *et al*: **A hereditary moyamoya syndrome with multisystemic manifestations**. *Neurology* 2010, **75**(3):259-264 [doi.org/10.1212/WNL.0b013e3181e8ee3f.](https://doi.org/10.1212/WNL.0b013e3181e8ee3f.)

530. Miskinyte S, Butler MG, Hervé D, Sarret C, Nicolino M, Petralia JD, Bergametti F, Arnould M, Pham VN, Gore AV *et al*: **Loss of BRCC3 deubiquitinating enzyme leads to abnormal angiogenesis and is associated with syndromic moyamoya**. *American journal of human genetics* 2011, **88**(6):718-728 [doi.org/10.1016/j.ajhg.2011.04.017.](https://doi.org/10.1016/j.ajhg.2011.04.017.)

531. Janczar S, Kosinska J, Ploski R, Pastorczak A, Wegner O, Zalewska-Szewczyk B, Paige AJ, Borowiec M, Mlynarski W: **Haemophilia A and cardiovascular morbidity in a female SHAM syndrome carrier due to skewed X chromosome inactivation**. *European journal of medical genetics* 2016, **59**(1):43-47 [doi.org/10.1016/j.ejmg.2015.12.004.](https://doi.org/10.1016/j.ejmg.2015.12.004.)

532. Jourdy Y, Chatron N, Fretigny M, Carage ML, Chambost H, Claeyssens-Donadel S, Roussel-Robert V, Negrier C, Sanlaville D, Vinciguerra C: **Molecular cytogenetic characterization of five F8 complex rearrangements: utility for haemophilia A genetic counselling**. *Haemophilia : the official journal of the World Federation of Hemophilia* 2017, **23**(4):e316-e323 [doi.org/10.1111/hae.13218.](https://doi.org/10.1111/hae.13218.)