**Table 1: Microdeletion and Microduplication Syndromes**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Name of MMS | Coordinates | Assembly | Reference(s) | hg19 Coordinates | Additional Notes: |
| 1p36 Syndrome | chr1:834083-6289973 | hg19 | DECIPHER | chr1:834083-6289973 | ClinGen Coordinates |
| chr1:10001-12780116 | GRCh38 | chr1:10001-12840259 | DECIPHER Coordinates |
| ATAD3 Cluster (1p36.33) | chr1:1385069-1470067 | hg19 | Desai\_2017[1](#_ENREF_1); Gunning\_2020[2](#_ENREF_2) | chr1:1385069-1470067 | Coordinates correspond to the ATAD3A-C gene cluster |
| 1p35 | chr1:28751378-33588455 | hg19 | Wilson\_2015[3](#_ENREF_3); Gennarino\_2018[4](#_ENREF_4) | chr1:28751378-33588455 | Coordinates correspond to Subject 1 |
| chr1:31404353-31538564 | hg19 | chr1:31404353-31538564 | *PUM1* Coordinates |
| 1p32.2-p31.3 | chr1:57633718-67087056 | GRCh38 | Lu\_2007[5](#_ENREF_5); Koehler\_2010[6](#_ENREF_6); Chen\_2011[7](#_ENREF_7); Ji\_2014[8](#_ENREF_8); Rao\_2014[9](#_ENREF_9); Kehrer\_2015[10](#_ENREF_10); Labonne\_2016[11](#_ENREF_11); Bayat\_2017[12](#_ENREF_12) | chr1:58099390-67552739 | Coordinates correspond to DGDP005 in Labonne *et al.* |
| chr1:61700460-62119733 | hg19 | chr1:61700460-62119733 | Coordinates correspond to DCP285169 in Labonne *et al.* |
| 1p21.3 | chr1:97500000-98720000 | hg18 | Carter\_2011[13](#_ENREF_13); Willemsen\_2011[14](#_ENREF_14); D’Angelo\_2018[15](#_ENREF_15) | chr1:97727412-98947412 | Coordinates correspond to SRO identified by Willemsen *et al.* |
| chr1:95696444-99846176 | hg19 | chr1:95696444-99846176 | Coordinates correspond to the start of Patient 7 to the end of Patient 6 in D’Angelo *et al.* |
| 1q21.1 TAR | chr1:145413192-145610627 | hg19 | Klopocki\_2007[16](#_ENREF_16); Brunetti-Pierri\_2008[17](#_ENREF_17); Mefford\_2008[18](#_ENREF_18); Rosenfeld\_2012[19](#_ENREF_19); Pang\_2020[20](#_ENREF_20) | chr1:145413192-145610627 | Coordinates correspond to the 200-kb TAR region defined in Klopocki *et al.* |
| chr1:145413192-145826450 | hg19 | chr1:145413192-145826450 | Coordinates correspond to the 500-kb TAR region defined in Klopocki *et al.* |
| 1q21.1 Neuro | chr1:146577486-147394506 | hg19 | Klopocki\_2007[16](#_ENREF_16); Brunetti-Pierri\_2008[17](#_ENREF_17); Mefford\_2008[18](#_ENREF_18); Rosenfeld\_2012[19](#_ENREF_19); Pang\_2020[20](#_ENREF_20) | chr1:146577486-147394506 | ClinGen Coordinates |
| chr1:147061832-148411223 | GRCh38 | chr1:146533376-147883376 | DECIPHER Coordinates |
| 1q21.1 TAR Neuro | chr1:145388978-147828089 | hg19 | Klopocki\_2007[16](#_ENREF_16); Brunetti-Pierri\_2008[17](#_ENREF_17); Mefford\_2008[18](#_ENREF_18); Rosenfeld\_2012[19](#_ENREF_19); Pang\_2020[20](#_ENREF_20) | chr1:145388978-147828089 | Coordinates correspond to the Class II region defined in Pang *et al.* |
| chr1:145118354-148026038 | hg19 | chr1:145118354-148026038 | Coordinates correspond to region between the proximal and distal SegDup cluster |
| 1q24-q25 Proximal | chr1:161650414-174926542 | hg19 | Burkardt\_2011[21](#_ENREF_21); Chatron\_2015[22](#_ENREF_22); Lam\_2016[23](#_ENREF_23);  Mackenroth\_2016[24](#_ENREF_24); Fichera\_2020[25](#_ENREF_25) | chr1:161650414-174926542 | Coordinates correspond to Patient 3 of Chatron *et al.* |
| chr1:164501003-167022133 | hg19 | chr1:164501003-167022133 | Coordinates correspond to the Proximal SRO defined by Chatron *et al.* |
| 1q24-q25 Intermediate | chr1:168556004-174427602 | hg19 | Burkardt\_2011[21](#_ENREF_21); Chatron\_2015[22](#_ENREF_22); Lam\_2016[23](#_ENREF_23);  Mackenroth\_2016[24](#_ENREF_24); Fichera\_2020[25](#_ENREF_25) | chr1:168556004-174427602 | Coordinates correspond to the region between the end of Case 1 of Fichera *et al.*  and the end of Patient 7 of Chatron *et al.* |
| 1q24-q25 Distal | chr1:170500104-184283033 | hg19 | Burkardt\_2011[21](#_ENREF_21); Chatron\_2015[22](#_ENREF_22); Lam\_2016[23](#_ENREF_23);  Mackenroth\_2016[24](#_ENREF_24); Fichera\_2020[25](#_ENREF_25) | chr1:170500104-184283033 | Coordinates correspond to Case 3 of Fichera *et al.* |
| chr1:178514910-181269712 | hg19 | chr1:178514910-181269712 | Coordinates correspond to the Distal SRO defined by Chatron *et al.* |
| 1q41-q42 | chr1:217316327-224182462 | hg18 | Rosenfeld\_2011[26](#_ENREF_26); Au\_2014[27](#_ENREF_27); Spreiz\_2014[28](#_ENREF_28);  Cassina\_2015[29](#_ENREF_29); Yanagishita\_2019[30](#_ENREF_30) | chr1:219249704-226115839 | Coordinates correspond to Subject 13 of Rosenfeld *et al.* |
| chr1:222988431-224622001 | hg19 | chr1:222988431-224622001 | Coordinates correspond to the region between genes: *DISP1* and *WDR26* |
| 1q43-q44 | chr1:239559031-245532863 | hg18 | Ballif\_2012[31](#_ENREF_31); Thierry\_2012[32](#_ENREF_32); Depienne\_2017[33](#_ENREF_33); Chen\_2020[34](#_ENREF_34) | chr1:241492408-247466240 | Coordinates correspond to Patient 3 |
| chr1:243287730-245318287 | hg19 | chr1:243287730-245318287 | ClinGen Coordinates |
|  |  |  |  |  |  |
| 2p25.3 (MYTIL) | chr2:0-3014104 | hg19 | Doco-Fenzy\_2014[35](#_ENREF_35); De Rocker\_2015[36](#_ENREF_36) | chr2:0-3014104 | Coordinates correspond to Rocker *et al.* Patient 1 |
| chr2:1792885-2335045 | hg19 | chr2:1792885-2335045 | *MYTIL* Coordinates |
| 2p21 | chr2:44183312-44362445 | GRCh38 | DECIPHER | chr2:44410451-44589584 | DECIPHER Coordinates |
| NRXN1 (2p16.3) | chr2:50145643-51259674 | hg19 | Béna\_2013[37](#_ENREF_37); Lowther\_2015[38](#_ENREF_38); Al Shehhi\_2019[39](#_ENREF_39) | chr2:50145643-51259674 | *NRXN1* Coordinates |
| chr2:50847740-51170975 | hg18 | chr2:50994236-51317471 | Coordinates correspond to Patient 10 of Béna *et al.* |
| 2p16.1-p15 | chr2:59139200-62488871 | hg19 | Kaminsky\_2011[40](#_ENREF_40); Piccione\_2012[41](#_ENREF_41);  Hancarova\_2013[42](#_ENREF_42); Mimouni-Bloch\_2015[43](#_ENREF_43) | chr2:59139200-62488871 | ClinGen Coordinates |
| 2p15-p14 | chr2:65490660-67666133 | hg19 | Kaminsky\_2011[40](#_ENREF_40); Wohlleber\_2011[44](#_ENREF_44); Hancarova\_2013[45](#_ENREF_45); Mathieu\_2017[46](#_ENREF_46); Pavone\_2019[47](#_ENREF_47) | chr2:65490660-67666133 | Coordinates correspond to Patient 2 of Matthieu *et al.* |
| 2p12-p11.2 | chr2:76854464-87287935 | hg17 | Barber\_2005[48](#_ENREF_48); Tzschach\_2009[49](#_ENREF_49); Writzl\_2009[50](#_ENREF_50) | chr2:76942809-87376277 | Coordinates correspond to the patient in Writzl *et al.* |
| 2q11.2 | chr2:96739309-97652262 | hg19 | Riley\_2015[51](#_ENREF_51); Wolfe\_2018[52](#_ENREF_52) | chr2:96739309-97652262 | Coordinates correspond to Subject #3 of Riley *et al.* |
| 2q13 | chr2:111449141-112746937 | hg19 | Riley\_2015[51](#_ENREF_51); Wolfe\_2018[52](#_ENREF_52) | chr2:111449141-112746937 | Coordinates correspond to the SRO defined by Wolfe *et al.* |
| Mowat Wilson (2q22.3) | chr2:145141942-145277958 | hg19 | Wilson\_2003[53](#_ENREF_53); Zweier\_2003[54](#_ENREF_54); Ishihara\_2004[55](#_ENREF_55); Ivanovski\_2018[56](#_ENREF_56) | chr2:145141942-145277958 | ClinGen Coordinates |
| 2q23.1  (MBD5) | chr2:148778580-149271046 | hg19 | Jaillard\_2009[57](#_ENREF_57); van Bon\_2010[58](#_ENREF_58); Williams\_2010[59](#_ENREF_59); Talkowski\_2011[60](#_ENREF_60) | chr2:148778580-149271046 | ClinGen Coordinates |
| chr2:147200000-150100000 | hg18 | chr2:147483530-150391754 | Coordinates correspond to Patient 4 of van Bon *et al.* |
| 2q31.1 | chr2:166519354-173745491 | hg18 | Cho\_2010[61](#_ENREF_61); Kantaputra\_2010[62](#_ENREF_62); Mitter\_2010[63](#_ENREF_63);  Theisen\_2010[64](#_ENREF_64); Dimitrov\_2011[65](#_ENREF_65); Ghoumid\_2011;  Gurrieri\_2013[66](#_ENREF_66); Sowińska-Seidler\_2014[67](#_ENREF_67) | chr2:166811108-174037245 | Coordinates correspond to Subject 2 of Theisen *et al.* |
| chr2:166293773-186068927 | hg18 | chr2:166585527-186360682 | Coordinates correspond to Patient 1 of Mitter *et al.* |
| chr2:175712131-183736599 | hg18 | chr2:176003885-184028354 | Coordinates correspond to Subject 10 of Theisen *et al.* |
| chr2:176957533-177055635 | hg19 | chr2:176957533-177055635 | Coordinates correspond to HOXD gene cluster, from *HOXD13* to *HOXD1* |
| 2q31.2-q32.3 | chr2:177644267-191382006 | hg18 | Prontera\_2009[68](#_ENREF_68); Cocchella\_2010[69](#_ENREF_69);  Manolakos\_2010[70](#_ENREF_70); Ferreira\_2012[71](#_ENREF_71) | chr2:177936021-191673761 | Coordinates correspond to the patient in Prontera *et al.* |
| chr2:182321619-185804214 | hg19 | chr2:182321619-185804214 | Coordinates correspond to the SRO defined by Ferreira *et al.* between: *IGTA4* and *ZNF804A* |
| 2q33.1 | chr2:196060397-204342216 | GRCh38 | Rosenfeld\_2009[72](#_ENREF_72); Balasubramanian\_2011[73](#_ENREF_73); Zarate\_2017[74](#_ENREF_74) | chr2:196925121-205206939 | DECIPHER Coordinates |
| chr2:200134223-200329831 | hg19 | chr2:200134223-200329831 | *SATB2* Coordinates |
| 2q35 | chr2:219919142-220025587 | hg19 | Klopocki\_2011[75](#_ENREF_75); Yuksel-Apak\_2012[76](#_ENREF_76);  Spielmann\_2013[77](#_ENREF_77); Barroso\_2015[78](#_ENREF_78) | chr2:219919142-220025587 | Coordinates correspond to the region between: *IHH* and *NHEJ1* |
| 2q37 | chr2:236402733-243199373 | hg19 | Williams\_2010[79](#_ENREF_79); Villavicencio-Lorini\_2013[80](#_ENREF_80); Wheeler\_2014[81](#_ENREF_81);  Jean-Marçais\_2015[82](#_ENREF_82); Le\_2019[83](#_ENREF_83) | chr2:236402733-243199373 | Coordinates correspond to the region between *AGAP1* and qter |
| chr2:239954693-242930600 | hg19 | chr2:239954693-242930600 | ClinGen Coordinates |
| chr2:239048167-239400949 | GRCh38 | chr2:239969863-240322643 | DECIPHER Coordinates |
|  |  |  |  |  |  |
| 3p25.3 | chr3:8922192-9885334 | hg19 | Gunnarsson\_2010[84](#_ENREF_84); Bownass\_2014[85](#_ENREF_85); Dikow\_2014[86](#_ENREF_86): Kuechler\_2014[87](#_ENREF_87) | chr3:8922192-9885334 | Coordinates correspond to the start of Bownass *et al.* and the end of Gunnarsson *et al.* |
| chr3:8856000-11305600 | hg19 | chr3:8856000-11305600 | Coordinates correspond to Patient 5 of Kuechler *et al.* |
| chr3:10391284-11516688 | hg19 | chr3:10391284-11516688 | Coordinates correspond to Patient 1 of Dikow *et al.* |
| 3p21.31 | chr3:48301437-51364610 | hg18 | Haldeman-Englert\_2009[88](#_ENREF_88); Eto\_2013[89](#_ENREF_89); Lovrecic\_2016[90](#_ENREF_90) | chr3:48326433-51389570 | Coordinates correspond to the Patient in Haldeman-Englert *et al.* |
| chr3:44251270-47936671 | hg19 | chr3:44251270-47936671 | Coordinates correspond to DECIPHER case 258176 from Lovrecic *et al.* |
| 3q13.31 | chr3:115335356-115916848 | hg18 | Molin\_2012[91](#_ENREF_91); Vuillaume\_2013[92](#_ENREF_92); Hervé\_2016[93](#_ENREF_93) | chr3:113852666-114434158 | Coordinates correspond to Case 13 and the SRO defined by Molin *et al.* |
| chr3:105911706-120983616 | hg18 | chr3:104429016-119500926 | Coordinates correspond to Case 5 of Molin *et al.* |
| chr3:112976429-120183473 | hg18 | chr3:111493740-118700783 | Coordinates correspond to Case 8 of Molin *et al.* |
| 3q22.1-q25.2 | chr3:137845310-156872233 | hg19 | Lim\_2011[94](#_ENREF_94); Moortgat\_2011[95](#_ENREF_95); Ferraris\_2013[96](#_ENREF_96);  Bertini\_2016[97](#_ENREF_97); Ramineni\_2016[98](#_ENREF_98) | chr3:137845310-156872233 | Coordinates correspond to the start of Lim *et al.* and the end of Bertini *et al*. Patient 6 |
| 3q27.3-q28 | chr3:185445864-192541120 | hg19 | Thevenon\_2014[99](#_ENREF_99); Ponzi\_2015[100](#_ENREF_100) | chr3:185445864-192541120 | Coordinates correspond to the start of Patient 1 to the end of Patient 2 of Thevenon *et al.* |
| chr3:186461680-187832449 | hg19 | chr3:186461680-187832449 | Coordinates correspond to SRO1 defined by Thevenon *et al.* |
| chr3:185816939-186461680 | hg19 | chr3:185816939-186461680 | Coordinates correspond to SRO2 defined by Thevenon *et al.* |
| chr3:188592241-190521638 | hg19 | chr3:188592241-190521638 | Coordinates correspond to the patients reported in Ponzi *et al.* |
| 3q29 | chr3:195999964-197617792 | GRCh38 | DECIPHER | chr3:195756054-197344662 | DECIPHER Coordinates |
|  |  |  |  |  |  |
| Wolf-Hirschhorn (4p16.3) | chr4:331568-2010962 | hg19 | Maas\_2008[101](#_ENREF_101); Ho\_2016[102](#_ENREF_102); Corrêa\_2018[103](#_ENREF_103) | chr4:331568-2010962 | ClinGen Coordinates |
| chr4:71552-3845097 | GRCh38 | chr4:71444-3846824 | Coordinates correspond to Case 1 of Corrêa *et al.* |
| chr4:68345-9785068 | hg19 | chr4:68345-9785068 | Coordinates correspond to Patient 15 of Ho *et al.* |
| 4q21 | chr4:76077557-85788996 | hg18 | Harada\_2002[104](#_ENREF_104); Bonnet\_2010[105](#_ENREF_105); Dukes-Rimsky\_2011[106](#_ENREF_106); Hu\_2017[107](#_ENREF_107) | chr4:75858534-85569972 | Coordinates correspond to Patient 6 of Bonnet *et al.* |
| chr4:82228875-83601083 | hg18 | chr4:82009852-83382059 | Coordinates correspond to the SRO defined by Bonnet *et al.* |
|  |  |  |  |  |  |
| Cri-du-chat (5p15) | chr5:10001-12533192 | GRCh38 | DECIPHER | chr5:10001-12533304 | DECIPHER Coordinates |
| 5p13 | chr5:36487985-37556112 | hg18 | Yan\_2009[108](#_ENREF_108); Romero\_2012[109](#_ENREF_109); Lucarelli\_2017[110](#_ENREF_110) | chr5:36452228-37520355 | Coordinates correspond to Patient 3 of Yan *et al.* |
| chr5:36845462-37417228 | hg18 | chr5:36809705-37381471 | Coordinates correspond to the start of Patient 1 to the end of Patient 5 of Yan *et al.* |
| 5q11.2 | chr5:50500000-56300000 | hg19 | Jaillard\_2011[111](#_ENREF_111); Lindstrand\_2014[112](#_ENREF_112); Snijders-Blok\_2014[113](#_ENREF_113);  Fontana\_2016[114](#_ENREF_114); Arora\_2019[115](#_ENREF_115) | chr5:50500000-56300000 | Coordinates correspond to Patient 2 of Snijders Blok *et al.* |
| 5q12 | chr5:56264851-62021754 | hg18 | Jaillard\_2011[111](#_ENREF_111); Lindstrand\_2014[112](#_ENREF_112); Fontana\_2016[114](#_ENREF_114);  Arora\_2019[115](#_ENREF_115); Cellamare\_2021[116](#_ENREF_116) | chr5:56229094-61985998 | Coordinates correspond to Patient 2 of Jaillard *et al.* |
| 5q14.3-q15 Proximal | chr5:86142512-90712814 | hg18 | Cardoso\_2009[117](#_ENREF_117); Engels\_2009[118](#_ENREF_118); Sobreira\_2009[119](#_ENREF_119);  Le Meur\_2010[120](#_ENREF_120); Hotz\_2013[121](#_ENREF_121) | chr5:86106756-90677058 | Coordinates correspond to Patient 6 of Le Meur *et al.* |
| chr5:88014058-88199922 | hg19 | chr5:88014058-88199922 | *MEF2C* Coordinates |
| 5q14.3-q15 Distal | chr5:90787099-98232469 | hg18 | Brown\_2009[122](#_ENREF_122); Cardoso\_2009[117](#_ENREF_117); Sobreira\_2009[119](#_ENREF_119);  Le Meur\_2010[120](#_ENREF_120); Al-Kateb\_2013[123](#_ENREF_123) | chr5:90751343-98204569 | Coordinates correspond to Sobreira *et al.* |
| chr5:92742875-92972632 | hg18 | chr5:92717119-92946876 | Coordinates correspond to SRO defined by Al-Kateb *et al.* |
| FAP (5q22.2) | chr5:112707504-112846239 | GRCh38 | DECIPHER | chr5:112043201-112181936 | DECIPHER Coordinates |
| ADLD (5q23.2) | chr5:126776622-126837020 | GRCh38 | DECIPHER | chr5:126112314-126172712 | DECIPHER Coordinates |
| 5q31.3 | chr5:139422959-139592820 | hg18 | Shimojima\_2011[124](#_ENREF_124); Hosoki\_2012[125](#_ENREF_125);  Brown\_2013[126](#_ENREF_126); Shimojima\_2018[127](#_ENREF_127) | chr5:139442775-139612636 | Coordinates correspond to start of Brown *et al* Patient 2 and end of Hosoki *et al.* Patient 3*.* |
| chr5:138011434-141309459 | hg18 | chr5:137983535-141329275 | Coordinates correspond to the start of Hosoki *et al.* Patient 3 and the end of Brown *et al.* Patient 2. |
| Sotos Syndrome (5q35) | chr5:176297633-177625115 | hg19 | DECIPHER | chr5:175724636-177052116 | DECIPHER Coordinates |
| chr5:176560080-176727214 | hg19 | chr5:176560080-176727214 | *NSD1* Coordinates |
|  |  |  |  |  |  |
| 6p25 | chr6:203619-2741187 | hg19 | Aldinger\_2009[128](#_ENREF_128); Delahaye\_2012[129](#_ENREF_129);  Vernon\_2013[130](#_ENREF_130); Linhares\_2015[131](#_ENREF_131) | chr6:203619-2741187 | Coordinates correspond to patient reported in Vernon *et al.* |
| chr6:485138-2245868 | hg19 | chr6:485138-2245868 | Coordinates correspond to the SRO defined by Linhares *et al.* |
| 6p25.1p24.3 | chr6:4745144-10384769 | hg19 | Kuipers\_2012[132](#_ENREF_132); Qi\_2015[133](#_ENREF_133); Kent\_2020[134](#_ENREF_134); Tassano\_2021[135](#_ENREF_135) | chr6:4745144-10384769 | Coordinates correspond to patient reported in Qi *et al.* |
| 6q12-q14.1 | chr6:64535801-80178353 | hg19 | Wang\_2009[136](#_ENREF_136); Engwerda\_2018[137](#_ENREF_137) | chr6:64535801-80178353 | Coordinates correspond to Patient 1 of Wang *et al.* |
| 6q13-q14.2 | chr6:72548959-84576295 | hg19 | Wang\_2009[136](#_ENREF_136); Engwerda\_2018[137](#_ENREF_137) | chr6:72548959-84576295 | Coordinates correspond to Patient 2 of Wang *et al.* |
| 6q14.1-q14.3 | chr6:80767339-88299283 | hg19 | Lowry\_2013[138](#_ENREF_138); Engwerda\_2018[137](#_ENREF_137) | chr6:80767339-88299283 | Coordinates correspond to Lowry *et al.* |
| 6q14.1-q15 | chr6:86137888-102703574 | hg19 | Dworschak\_2015[139](#_ENREF_139); Engwerda\_2018[137](#_ENREF_137) | chr6:86137888-102703574 | Coordinates correspond to Patient 1 of Dworschak *et al.* |
| 6q14.3-q16.3 | chr6:72100590-97078142 | hg19 | Van Esch\_2010[140](#_ENREF_140); Engwerda\_2018[137](#_ENREF_137) | chr6:72100590-97078142 | Coordinates correspond to Patient 3 of Van Esch *et al.* |
| 6q24-q25 | chr6:143740000-152480000 | hg18 | Thienpont\_2010[141](#_ENREF_141) | chr6:143698307-152438307 | Coordinates correspond to the start of Patient A to the end of Patient B of Thienpont *et al.* |
| chr6:149230000-149960000 | hg18 | chr6:149188307-149918307 | Coordinates correspond to the SRO defined by Thienpont *et al.* |
| 6q25 | chr6:156550000-157790000 | hg18 | Nagamani\_2009[142](#_ENREF_142); Santen\_2014[143](#_ENREF_143); Ronzoni\_2016[144](#_ENREF_144); Zhong\_2021[145](#_ENREF_145) | chr6:156508308-157870012 | Coordinates correspond to the end of Patient C to the end of Patient J of Thienpoint *et al.* |
| chr6:152307705-160422834 | hg19 | chr6:152307705-160422834 | Coordinates correspond to Patient of Zhong *et al.* |
|  |  |  |  |  |  |
| 7p22.1 | chr7:3483223-7325864 | hg19 | Caselli\_2015[146](#_ENREF_146); Shimojima\_2016[147](#_ENREF_147) | chr7:3483223-7325864 | Coordinates correspond to DECIPHER Case 249672 referenced in Caselli *et al.* |
| chr7:5492967-5864851 | hg19 | chr7:5492967-5864851 | Coordinates correspond to Patient 4,5 of Shimojima *et al.* |
| WBS (7q11.23) | chr7:72744455-74142510 | hg19 | Del Campo\_2006[148](#_ENREF_148); Van der Aa\_2009[149](#_ENREF_149); Ramocki\_2010[150](#_ENREF_150); Faundes\_2012[151](#_ENREF_151); Porter\_2012[152](#_ENREF_152) | chr7:72744455-74142510 | Coordinates correspond to the 1.5 Mb Common Region; ClinGen Coordinates |
| chr7:72716514-74565623 | hg19 | chr7:72716514-74565623 | Coordinates correspond to the 1.8 Mb Region; NSUN5 to GTF2IRD2B |
| chr7:72716514-76256141 | hg19 | chr7:72716514-76256141 | Coordinates correspond to the entire WBS Region (LCR-P to LCR-D) |
| WBS Distal (7q11.23) | chr7:75158048-76063176 | hg19 | Komoike\_2010[153](#_ENREF_153); Ramocki\_2010[150](#_ENREF_150) | chr7:75158048-76063176 | ClinGen Coordinates |
| SHFM1 (7q21.3) | chr7:95401818-96654143 | hg19 | Velinov\_2012[154](#_ENREF_154); Rattanasopha\_2014[155](#_ENREF_155); Sowińska-Seidler\_2014[67](#_ENREF_67); Tayebi\_2014[156](#_ENREF_156); Delgado\_2015[157](#_ENREF_157); Rasmussen\_2016[158](#_ENREF_158) | chr7:95401818-96654143 | Coordinates correspond to the region between *DYNC1I1* and *DLX5* |
|  |  |  |  |  |  |
| 8p23.1 | chr8:8242533-11907120 | GRCh38 | DECIPHER | chr8:8100055-11764629 | DECIPHER Coordinates |
| 8q12 | chr8:60219746-64072039 | hg19 | Lehman\_2009[159](#_ENREF_159); Amouroux\_2012[160](#_ENREF_160); Luo\_2012[161](#_ENREF_161);  Baroncini\_2013[162](#_ENREF_162); Palumbo\_2013[163](#_ENREF_163) | chr8:60219746-64072039 | Coordinates correspond to the patient reported in Amouroux *et al.* |
| 8q13 | chr8:69900100-72552212 | hg19 | Isidor\_2009[164](#_ENREF_164); Isidor\_2010[165](#_ENREF_165); Hamilton\_2016[166](#_ENREF_166) | chr8:69900100-72552212 | Coordinates correspond to Patient in Hamilton *et al.* |
| chr8:70378859-70745404 | hg19 | chr8:70378859-70745404 | Coordinates correspond to the region between: *SULF1* and *SLCO5A1* |
| 8q21.11 | chr8:74915253-81924253 | hg19 | Palomares\_2011[167](#_ENREF_167); Niyazov\_2015[168](#_ENREF_168); Quintela\_2015[169](#_ENREF_169); Happ\_2016[170](#_ENREF_170) | chr8:74915253-81924253 | Coordinates correspond to the Patient in Quintela *et al.* |
| chr8:76314229-76854003 | GRCh38 | chr8:77226464-77766239 | DECIPHER Coordinates |
| 8q22.1 | chr8:94358695-96281462 | hg19 | Raas-Rothschild\_2009[171](#_ENREF_171); Jain\_2010[172](#_ENREF_172); Allanson\_2012[173](#_ENREF_173); Overhoff\_2014[174](#_ENREF_174); Jamuar\_2015[175](#_ENREF_175) | chr8:94358695-96281462 | Coordinates correspond to the Critical Region in Overhoff *et al.* between genes:  LINC00535 and C8orf37 |
| chr8:84974143-96085410 | hg19 | chr8:84974143-96085410 | Coordinates correspond to patients without NMLFS; Patient 9 of Allanson *et al.* |
| 8q22.2–q22.3 | chr8:102017156-103939573 | hg18 | Kuechler\_2011[176](#_ENREF_176); Kuroda\_2014[177](#_ENREF_177); Sinajon\_2015[178](#_ENREF_178) | chr8:101947980-103870397 | Coordinates correspond to Patient 5 of Kuechler *et al.* |
| chr8:100025224-106469347 | hg18 | chr8:99956048-106400171 | Coordinates correspond to Patient 4 of Kuechler *et al.* |
| 8q23.1-q24.12 | chr8:116420724-119124058 | hg19 | Pereza\_2012[179](#_ENREF_179); Li\_2015[180](#_ENREF_180); Schinzel\_2013[181](#_ENREF_181); Selenti\_2015[182](#_ENREF_182) | chr8:116420724-119124058 | Coordinates correspond to the minimal region between *TRPS1* and *EXT1* |
| chr8:114338391-121241253 | hg19 | chr8:114338391-121241253 | Coordinates correspond to the start of Li *et al.* to the end of Patient 3 of Schinzel *et al.* |
| 8q24.3 | chr8:144873090-144911556 | hg19 | Dauber\_2013[183](#_ENREF_183); Wells\_2016[184](#_ENREF_184); Abdin\_2019[185](#_ENREF_185) | chr8:144873090-144911556 | Coordinates correspond to the region between *SCRIB* and *PUF60* |
| chr8:144159121-145573790 | hg19 | chr8:144159121-145573790 | Coordinates correspond to Patient 5 of Dauber *et al.* |
|  |  |  |  |  |  |
| SRXY4 (9p24.3) | chr9:1-1057554 | hg19 | Barbaro\_2009[186](#_ENREF_186); Tannour-Louet\_2010[187](#_ENREF_187);  Onesimo\_2012[188](#_ENREF_188); Quinonez\_2013[189](#_ENREF_189) | chr9:1-1057554 | Coordinates correspond to p-terminal to *DMRT2* |
| 9p Terminal | chr9:1-4300035 | hg19 | Barbaro\_2009[186](#_ENREF_186); Hauge\_2008[190](#_ENREF_190); Swinkels\_2008[191](#_ENREF_191); Onesimo\_2012[188](#_ENREF_188); Mitsui\_2013[192](#_ENREF_192); Quinonez\_2013[189](#_ENREF_189) | chr9:1-4300035 | Coordinates correspond to p-terminal to *GLIS3* |
| chr9:1-14734664 | hg19 | chr9:1-14734664 | Coordinates correspond to Patients 1-2 in Vissers *et al.* |
| chr9:8053379-16077945 | hg19 | chr9:8053379-16077945 | Coordinates correspond to the critical region defined by Christ *et al.* between D9S286, D9S285 |
| 9p13.3-p13.1 | chr9:33809009-38653271 | hg18 | Niemi\_2012[193](#_ENREF_193); Crone\_2016[194](#_ENREF_194); Ferreira\_2020[195](#_ENREF_195) | chr9:33819009-38663271 | Coordinates correspond to Patient 2 of Niemi *et al.* |
| 9q22.3 | chr9:90333007-101185240 | hg18 | Shimojima\_2009[196](#_ENREF_196); Muller\_2012[197](#_ENREF_197); Reichert\_2015[198](#_ENREF_198); Yamada\_2020[199](#_ENREF_199); Ewing\_2021[200](#_ENREF_200) | chr9:91143187-102145419 | Coordinates correspond to Patient 2 of Muller *et al.* |
| chr9:95885026-98218797 | hg18 | chr9:96845205-99178976 | Coordinates correspond to patient reported in Shimojima *et al.* |
| chr9:98205264-98269699 | hg19 | chr9:98205264-98269699 | *PTCH1* Coordinates |
| 9q31.1-q31.3 | chr9:107945742-114439602 | hg19 | Mucciolo\_2014[201](#_ENREF_201); Nevado\_2014[202](#_ENREF_202) | chr9:107945742-114439602 | Coordinates correspond to Patient 2 reported in Mucciolo *et al.* |
| Kleefstra Syndrome (9q34.3) | chr9:137618991-137836126 | GRCh38 | DECIPHER | chr9:140513444-140730579 | DECIPHER Coordinates |
|  |  |  |  |  |  |
| 10p Terminal | chr10:0-10550771 | hg19 | Schuffenhauer\_1998[203](#_ENREF_203); Lichtner\_2000[204](#_ENREF_204);  Skrypnyk\_2002[205](#_ENREF_205); Yatsenko\_2004[206](#_ENREF_206); Lindstrand\_2010[207](#_ENREF_207); Melis\_2012[208](#_ENREF_208); Kim\_2017[209](#_ENREF_209); Shao\_2021[210](#_ENREF_210) | chr10:0-10550771 | Coordinates correspond to Patient "DW" in Lichtner *et al.* |
| 10p15.3 | chr10:101955-739804 | hg18 | DeScipio\_2012[211](#_ENREF_211); Poluha\_2017[212](#_ENREF_212); Shao\_2021[210](#_ENREF_210) | chr10:111955-749804 | Coordinates correspond to Patient 4 of DeScipio *et al.* |
| chr10:126361-3090934 | hg18 | chr10:136361-3100934 | Coordinates correspond to Patient 8 of DeScipio *et al.* |
| 10p14-p12 | chr10:8212176-19608181 | hg19 | Schuffenhauer\_1998[203](#_ENREF_203); Lichtner\_2000[204](#_ENREF_204);  Skrypnyk\_2002[205](#_ENREF_205); Yatsenko\_2004[206](#_ENREF_206); Lindstrand\_2010[207](#_ENREF_207); Melis\_2012[208](#_ENREF_208); Kim\_2017[209](#_ENREF_209); Shao\_2021[210](#_ENREF_210) | chr10:8212176-19608181 | Coordinates correspond to the start of Patient "CH92-304" from Yatsenko *et al.*  to the end of Patient "MEG" in Schuffenhauer *et al.* |
| HDR (10p14) | chr10:6721874-8919127 | hg19 | Schuffenhauer\_1998[203](#_ENREF_203); Lichtner\_2000[204](#_ENREF_204);  van Esch\_2000[213](#_ENREF_213); Fukami\_2011[214](#_ENREF_214); Shao\_2021[210](#_ENREF_210) | chr10:6721874-8919127 | Coordinates correspond to the region between the makers D10S189 to D10S226 |
| chr10:8096667-8117164 | hg19 | chr10:8096667-8117164 | Coordinates correspond to *GATA3* |
| DGS2 (10p14) | chr10:10550410-11253972 | hg19 | Schuffenhauer\_1998[203](#_ENREF_203); Lichtner\_2000[204](#_ENREF_204);  Lichtner\_2002[215](#_ENREF_215); Kim\_2017[209](#_ENREF_209); Shao\_2021[210](#_ENREF_210) | chr10:10550410-11253972 | Coordinates correspond to the region between the makers D10S547 and D10S585 |
| 10q22-q23 Syndrome | chr10:81682843-88739388 | hg19 | Alliman\_2010[216](#_ENREF_216); van Bon\_2011[217](#_ENREF_217) | chr10:81682843-88739388 | ClinGen Coordinates |
| SHFM3 (10q24) | chr10:102969344-103498069 | hg19 | Dimitrov\_2010[218](#_ENREF_218); Sowińska-Seidler\_2014[67](#_ENREF_67); Holder-Espinasse\_2019[219](#_ENREF_219) | chr10:102969344-103498069 | Coordinates correspond to Family 2 of Dimitrov *et al.* |
| Interstitial 10q26 | chr10:118506755-125492105 | hg19 | Choucair\_2015[220](#_ENREF_220); Faria\_2016[221](#_ENREF_221); Lin\_2016[222](#_ENREF_222) | chr10:118506755-125492105 | Coordinates correspond to Miller Patient 4 from Faria *et al.* |
| Terminal 10q26 | chr10:121385399-135534747 | hg19 | Yatsenko\_2009[223](#_ENREF_223); Faria\_2016[221](#_ENREF_221); Lin\_2016[222](#_ENREF_222) | chr10:121385399-135534747 | Coordinates correspond to Patient 1 of Lin *et al.* |
| chr10:128113575-135534747 | hg19 | chr10:128113575-135534747 | Coordinates correspond to *C10orf90* to qter |
|  |  |  |  |  |  |
| Beckwith-Wiedemann / Russell-Silver (11p15.5) | chr11:2016406-2906995 | hg19 | Chiesa\_2012[224](#_ENREF_224) | chr11:2016406-2906995 | Coordinates correspond to the region between *H19* and *CDKN1C* |
| ELP4 (11p13) | chr11:31531297-31805329 | hg19 | Addis\_2015[225](#_ENREF_225) | chr11:31531297-31805329 | Coordinates correspond to *ELP4* |
| WAGRO (11p13) | chr11:27676442-32457081 | hg19 | Han\_2008[226](#_ENREF_226); Han\_2013[227](#_ENREF_227); Ferreira\_2019[228](#_ENREF_228); Hall\_2019[229](#_ENREF_229) | chr11:27676442-32457081 | Coordinates correspond to the region between *BDNF* and WAGR Critical Region (*PAX6* and *WT1*) |
| WAGR (11p13) | chr11:31803509-32510988 | hg19 | Fischbach\_2005[230](#_ENREF_230); Han\_2008[226](#_ENREF_226); Han\_2013[227](#_ENREF_227);  Yamamoto\_2014[231](#_ENREF_231); Hall\_2019[229](#_ENREF_229) | chr11:31803509-32510988 | ClinGen Coordinates |
| Potocki-Shaffer  (11p11.2) | chr11:43894800-46152450 | hg19 | Swarr\_2010[232](#_ENREF_232); Kim\_2012[233](#_ENREF_233); Kim\_2019[234](#_ENREF_234); Trajkova\_2020[235](#_ENREF_235) | chr11:43894800-46152450 | ClinGen Coordinates |
| chr11:42312866-50419631 | GRCh38 | chr11:42334416-50378802 | Coordinates correspond to DECIPHER Case 286390 referenced in Tajkova *et al.* |
| 11q13 | chr11:67763646-71236931 | hg19 | Gregory-Evans\_2007[236](#_ENREF_236); Alsmadi\_2009[237](#_ENREF_237);  Ziebart\_2013[238](#_ENREF_238); Kim\_2016[239](#_ENREF_239) | chr11:67763646-71236931 | ClinGen Coordinates |
| chr11:69246102-69600647 | hg18 | chr11:69536921-69922999 | Coordinates correspond to patient reported in Ziebart *et al.* |
| chr11:69624736-69634192 | hg19 | chr11:69624736-69634192 | *FGF3* Coordinates |
| Jacobsen (11q23-qter) | chr11:119634290-135006516 | hg19 | Grossfeld\_2004[240](#_ENREF_240); Coldren\_2009[241](#_ENREF_241); Ji\_2010[242](#_ENREF_242); Ye\_2010[243](#_ENREF_243) | chr11:119634290-135006516 | Coordinates correspond to Phenotype Group ‘A’ of Grossfeld *et al.* |
| chr11:122297785-135006516 | hg19 | chr11:122297785-135006516 | Coordinates correspond to Phenotype Group ‘B’ of Grossfeld *et al.* |
| chr11:125086881-135006516 | hg19 | chr11:125086881-135006516 | Coordinates correspond to Phenotype Group ‘C’ of Grossfeld *et al.* |
| chr11:126225789-135006516 | hg19 | chr11:126225789-135006516 | Coordinates correspond to Phenotype Group ‘D’ of Grossfeld *et al.* |
| chr11:128142005-135006516 | hg19 | chr11:128142005-135006516 | Coordinates correspond to Phenotype Group ‘E’ of Grossfeld *et al.* |
|  |  |  |  |  |  |
| 12p13.33 | chr12:0-2537524 | hg18 | Rooryck\_2009[244](#_ENREF_244); Thevanon\_2013[245](#_ENREF_245); Silva\_2014[246](#_ENREF_246); Mio\_2020[247](#_ENREF_247) | chr12:0-2667263 | Coordinates correspond to the start of chr12 to the end of Thevanon P8 |
| 12q14 | chr12:55704299-58779521 | hg18 | Menten\_2007[248](#_ENREF_248); Buysse\_2009[249](#_ENREF_249); Lynch\_2011[250](#_ENREF_250);  Fischetto\_2017[251](#_ENREF_251); Dória\_2020[252](#_ENREF_252) | chr12:57418032-60493254 | Coordinates correspond to Case 3 of Lynch *et al.* |
| chr12:56192860-66309484 | hg18 | chr12:57906593-68023217 | Coordinates correspond to Case 1 of Lynch *et al.* |
| chr12:63342399-66780345 | hg18 | chr12:65056132-68494078 | Coordinates correspond to Patient 3 of Menten *et al.* |
| PACD (12q21.33) | chr12:91311800-91573359 | hg19 | Aldave\_2010[253](#_ENREF_253); Kim\_2014[254](#_ENREF_254); Odent\_2017[255](#_ENREF_255) | chr12:91311800-91573359 | Coordinates correspond to the SRO between the genes: *LINC00615* and *DCN* |
| 12q24.3 | chr12:121639241-123000639 | hg19 | Baple\_2010[256](#_ENREF_256); Qiao\_2013[257](#_ENREF_257); Chouery\_2013[258](#_ENREF_258);  Palumbo\_2015[259](#_ENREF_259); Labonne\_2016[260](#_ENREF_260) | chr12:121639241-123000639 | Coordinates correspond to the midpoint of Chouery *et al.* to the midpoint of Qiao *et al.* |
|  |  |  |  |  |  |
| 13q12.11-q12.13 | chr13:19498791-21439928 | hg18 | Der Kaloustian\_2011[261](#_ENREF_261); Tanteles\_2011[262](#_ENREF_262); Lagou\_2014[263](#_ENREF_263); Pavone\_2014[264](#_ENREF_264); Tominaga\_2019[265](#_ENREF_265) | chr13:20600792-22541928 | Coordinates correspond to the patient reported in Der Kaloustian *et al.* |
| chr13:20427493-26037670 | hg19 | chr13:20427493-26037670 | Coordinates correspond to Pavone *et al.* |
| chr13:23566962-24890143 | hg19 | chr13:23566962-24890143 | Coordinates correspond to DECIPHER Case #272677 from Pavone *et al.* |
| 13q12.3 | chr13:30768420-32166016 | hg19 | Bartholdi\_2014[266](#_ENREF_266); Mandrile\_2014[267](#_ENREF_267); D’Angelo\_2018[15](#_ENREF_15);  Wang\_2020[268](#_ENREF_268); Uguen\_2021[269](#_ENREF_269) | chr13:30768420-32166016 | Coordinates correspond to Patient 2 of Bartholdi *et al.* |
| chr13:30776767-31191510 | hg19 | chr13:30776767-31191510 | Coordinates correspond to the critical region between genes: *KATNAL1* and *HMGB1* |
| 13q14 | chr13:36383200-74000000 | hg18 | Caselli\_2007[270](#_ENREF_270); Mitter\_2011[271](#_ENREF_271); Castéra\_2013[272](#_ENREF_272) | chr13:37485200-75101999 | Coordinates correspond to Patient 12 of Castera *et al.* |
| chr13:40733729-60193507 | hg18 | chr13:41835729-61295506 | Coordinates correspond to Patient 9 of Mitter *et al.* |
| chr13:47072184-49032229 | hg18 | chr13:48174183-50134228 | Coordinates correspond to Patient 52 of Mitter *et al.* |
| chr13:48877883-49056026 | hg19 | chr13:48877883-49056026 | Coordinates correspond to *RB1* |
| Feingold Syndrome 2  (13q31.3) | chr13:90653738-92490593 | hg19 | de Pontual\_2011[273](#_ENREF_273); Ganjavi\_2014[274](#_ENREF_274);  Valdes-Miranda\_2014[275](#_ENREF_275); Muriello\_2019[276](#_ENREF_276) | chr13:90653738-92490593 | Coordinates correspond to the start of Patient 2 of Muriello *et al.* |
| 13q32.1 | chr13:95241606-95273292 | hg19 | Fares-Taie\_2015[277](#_ENREF_277); Al-Owaid\_2019[278](#_ENREF_278); Pozza\_2020[279](#_ENREF_279); Angée\_2021[280](#_ENREF_280) | chr13:95241606-95273292 | Coordinates correspond to the deletion SRO defined by Angée *et al.* |
| chr13:95119866-95438104 | hg19 | chr13:95119866-95438104 | Coordinates correspond to the duplication SRO defined by Angée *et al.* |
| 13q33-q34 | chr13:104753418-115107733 | hg19 | Kirchhoff\_2009[281](#_ENREF_281); Sagi-Dain\_2019[282](#_ENREF_282) | chr13:104753418-115107733 | Coordinates correspond to Patient 4 of Sagi-Dain *et al.* |
| chr13:109149281-115107733 | hg19 | chr13:109149281-115107733 | Coordinates correspond to Patient 8 of Sagi-Dain *et al.* |
|  |  |  |  |  |  |
| 14q11-q22 | chr14:26972703-38533633 | hg18 | Shimojima\_2009[283](#_ENREF_283); Torgyekes\_2011[284](#_ENREF_284); Fonseca\_2012[285](#_ENREF_285); Santen\_2012[286](#_ENREF_286); Gentile\_2016[287](#_ENREF_287) | chr14:27902863-39463882 | Coordinates correspond to Patient 2 of Torgyekes *et al.* |
| chr14:35443407-39527387 | hg19 | chr14:35443407-39527387 | Coordinates correspond to patient reported in Gentile *et al.* |
| chr14:27932292-33935994 | hg18 | chr14:28862541-34866243 | Coordinates correspond to Patient 1 of Torgyekes *et al.* |
| Frias Syndrome  (14q22-q23) | chr14:53001803-55443425 | hg19 | Brisset\_2014[288](#_ENREF_288); Martínez-Fernández\_2014[289](#_ENREF_289); Blackburn\_2019[290](#_ENREF_290) | chr14:53001803-55443425 | Coordinates correspond to Patient 2 of Blackburn *et al.* |
| chr14:54416455-54421270 | hg19 | chr14:54416455-54421270 | Coordinates correspond to *BMP4* |
| Temple / Kagami-Ogata  (14q32.2) | chr14:100394594-101504529 | hg19 | Kagami\_2005[291](#_ENREF_291); Béna\_2010[292](#_ENREF_292); Zada\_2014[293](#_ENREF_293); Severi\_2015[294](#_ENREF_294); Kagami\_2017[295](#_ENREF_295); Prasasya\_2020[296](#_ENREF_296) | chr14:100394594-101504529 | ClinGen Coordinates |
| chr14:101191391-101294616 | hg19 | chr14:101191391-101294616 | ClinGen Coordinates |
|  |  |  |  |  |  |
| Angelman / Prader-Willi BP1-3  (15q11.2-q13.1) | chr15:22832519-28379874 | hg19 | Christian\_1995[297](#_ENREF_297); Tan\_2011[298](#_ENREF_298); Kim\_2012[299](#_ENREF_299); Cox\_2015[300](#_ENREF_300) | chr15:22832519-28379874 | ClinGen Coordinates |
| 15q11.2 BP1-2 | chr15:22832519-23747996 | hg19 | Burnside\_2011[301](#_ENREF_301); von der Lippe\_2011[302](#_ENREF_302); Sahoo\_2011[303](#_ENREF_303) | chr15:22832519-23747996 | ClinGen Coordinates |
| chr15:22832519-23090897 | hg19 | chr15:22832519-23090897 | ClinGen Coordinates |
| Angelman / Prader-Willi BP2-3  (15q11.2-q13.1) | chr15:23747996-28379874 | hg19 | Christian\_1995[297](#_ENREF_297); Kaminsky\_2011[40](#_ENREF_40);  Tan\_2011[298](#_ENREF_298); Kim\_2012[299](#_ENREF_299); Cox\_2015[300](#_ENREF_300) | chr15:23747996-28379874 | ClinGen Coordinates |
| 15q13.1-q13.2 BP3-4 | chr15:28379874-31192889 | hg19 | Sharp\_2008[304](#_ENREF_304); van Bon\_2009[305](#_ENREF_305); Rosenfeld\_2011[306](#_ENREF_306) | chr15:28379874-31192889 | ClinGen Coordinates |
| 15q13.1-13.3 BP3-5 | chr15:28379874-32445405 | hg19 | Sharp\_2008[304](#_ENREF_304); van Bon\_2009[305](#_ENREF_305) | chr15:28379874-32445405 | ClinGen Coordinates |
| 15q13.3 BP4-5 | chr15:31192889-32445405 | hg19 | Miller\_2009[307](#_ENREF_307); van Bon\_2009[305](#_ENREF_305); Kaminsky\_2011[40](#_ENREF_40);  Sahoo\_2011[303](#_ENREF_303); Lowther\_2015[308](#_ENREF_308); Uddin\_2018[309](#_ENREF_309) | chr15:31192889-32445405 | ClinGen Coordinates |
| CHRNA7 (15q13.3) | chr15:32019621-32445405 | hg19 | Sahoo\_2011[303](#_ENREF_303); Hoppman-Chaney\_2013[310](#_ENREF_310);  Lowther\_2015[308](#_ENREF_308); Uddin\_2018[309](#_ENREF_309) | chr15:32019621-32445405 | ClinGen Coordinates |
| MEIS2 (15q14) | chr15:33894032-38659166 | hg19 | Johansson\_2014[311](#_ENREF_311); Chen\_2016[312](#_ENREF_312); Gambin\_2017[313](#_ENREF_313); Shimojima\_2017[314](#_ENREF_314) | chr15:33894032-38659166 | Coordinates correspond to Patient 3 of Gambin *et al.* |
| chr15:36790702-37404359 | hg19 | chr15:36790702-37404359 | Coordinates correspond to Patient 6 of Gambin *et al.* |
| 15q15.3 | chr15:43891761-43941039 | hg19 | Zhang\_2007[315](#_ENREF_315); Knijnenburg\_2009[316](#_ENREF_316); Vona\_2015[317](#_ENREF_317) | chr15:43891761-43941039 | Coordinates correspond to the region between genes: *STRC* and *CATSPER2* |
| FBN1 (15q21.1) | chr15:46098636-47145902 | hg18 | Mátyás\_2007[318](#_ENREF_318); Furtado\_2011[319](#_ENREF_319);  Hilhorst-Hofstee\_2011[320](#_ENREF_320); Dordoni\_2017[321](#_ENREF_321) | chr15:48311344-49358610 | Coordinates correspond to Patient 7 of Hilhorst-Hofstee *et al.* |
| chr15:48700503-48937985 | hg19 | chr15:48700503-48937985 | *FBN1* Coordinates |
| 15q24 AB | chr15:72963970-74343898 | hg19 | Magoulas\_2012[322](#_ENREF_322); Huynh\_2018[323](#_ENREF_323); Liu\_2020[324](#_ENREF_324) | chr15:72963970-74343898 | Coordinate correspond to patient reported in Huynh *et al.* |
| 15q24 AC | chr15:72963715-75508312 | hg19 | El-Hattab\_2009[325](#_ENREF_325); El-Hattab\_2010[326](#_ENREF_326); Magoulas\_2012[322](#_ENREF_322); Liu\_2020[324](#_ENREF_324) | chr15:72963715-75508312 | ClinGen Coordinates |
| 15q24 AD | chr15:72963715-75972909 | hg19 | El-Hattab\_2009[325](#_ENREF_325); Magoulas\_2012[322](#_ENREF_322); Mefford\_2012[327](#_ENREF_327); Liu\_2020[324](#_ENREF_324) | chr15:72963715-75972909 | ClinGen Coordinates |
| 15q24 BD | chr15:74120302-75680570 | GRCh38 | El-Hattab\_2009[325](#_ENREF_325); El-Hattab\_2010[326](#_ENREF_326);  Magoulas\_2012[322](#_ENREF_322); Mefford\_2012[327](#_ENREF_327); Liu\_2020[324](#_ENREF_324) | chr15:74412643-75972911 | DECIPHER Coordinates |
| 15q24 BE | chr15:72252000-75937000 | hg18 | El-Hattab\_2009[325](#_ENREF_325); Magoulas\_2012[322](#_ENREF_322); Mefford\_2012[327](#_ENREF_327); Liu\_2020[324](#_ENREF_324) | chr15:74464947-78149945 | Coordinates correspond to Patients 1,2 of El-Hattab *et al.* |
| Witteveen-Kolk Syndrome  (15q24 CD) | chr15:75631787-75972909 | hg19 | Witteveen\_2016[328](#_ENREF_328) | chr15:75631787-75972909 | ClinGen Coordinates |
| chr15:75661720-75744087 | hg19 | chr15:75661720-75744087 | Coordinates correspond to *SIN3A* |
| 15q24 DE | chr15:73858160-75969880 | hg18 | El-Hattab\_2009[325](#_ENREF_325) | chr15:76071105-78182825 | Coordinates correspond to Patients 6,7 of El-Hattab *et al.* |
| 15q25 | chr15:83213988-84714733 | hg19 | Wat\_2010[329](#_ENREF_329); Palumbo\_2012[330](#_ENREF_330); Doelken\_2013[331](#_ENREF_331) | chr15:83213988-84714733 | ClinGen Coordinates |
| chr15:85139652-85713001 | hg19 | chr15:85139652-85713001 | ClinGen Coordinates |
| chr15:83213988-85713001 | hg19 | chr15:83213988-85713001 | ClinGen Coordinates |
| 15q26 | chr15:98814741-101981189 | GRCh38 | Tatton-Brown\_2009[332](#_ENREF_332); Levy\_2012[333](#_ENREF_333); Poot\_2013[334](#_ENREF_334);  Leffler\_2016[335](#_ENREF_335); Cannarella\_2017 | chr15:99357970-102521392 | DECIPHER Coordinates |
| chr15:84151101-100276767 | hg18 | chr15:86350098-102459244 | Coordinates correspond to Patient 3 of Levy *et al.* |
|  |  |  |  |  |  |
| ATR-16 (16p13.3) | chr16:10001-784372 | GRCh38 | DECIPHER | chr16:60001-834372 | DECIPHER Coordinates |
| 16p13.3 | chr16:2097990-2185899 | hg19 | Oyazato\_2011[336](#_ENREF_336); Osumi\_2020[337](#_ENREF_337) | chr16:2097990-2185899 | Coordinates correspond to the genes: *TSC2* and *PKD1* |
| Rubinstein-Taybi (16p13.3) | chr16:3725054-3880120 | GRCh38 | DECIPHER | chr16:3775055-3930121 | DECIPHER Coordinates |
| Hao-Fountain Syndrome (16p13.2) | chr16:5486454-9203355 | hg19 | Hao\_2015[338](#_ENREF_338); Fountain\_2019[339](#_ENREF_339) | chr16:5486454-9203355 | Coordinates correspond to Patient 4 of Hao *et al.* |
| chr16:8969780-9211161 | hg19 | chr16:8969780-9211161 | Coordinates correspond to Patient 7 of Fountain *et al.* |
| chr16:8985951-9030543 | hg19 | chr16:8985951-9030543 | *USP7* Coordinates |
| 16p13.11 BP I-II | chr16:14892827-16392827 | GRCh38 | Ullmann\_2007[340](#_ENREF_340); Hannes\_2008[341](#_ENREF_341); de Kovel\_2010[342](#_ENREF_342); Ingason\_2011[343](#_ENREF_343) | chr16:14986684-16486684 | DECIPHER Coordinates |
| 16p13.11 BP I-III | chr16:14825307-18057186 | hg18 | Ullmann\_2007[340](#_ENREF_340); Hannes\_2008[341](#_ENREF_341); de Kovel\_2010[342](#_ENREF_342); Ingason\_2011[343](#_ENREF_343) | chr16:14917807-18149685 | Coordinates correspond to Patient 4826 of Kaminsky *et al.* |
| 16p13.11 | chr16:15511711-16292265 | hg19 | Ullmann\_2007[340](#_ENREF_340); Hannes\_2008[341](#_ENREF_341); de Kovel\_2010[342](#_ENREF_342); Ingason\_2011[343](#_ENREF_343) | chr16:15511711-16292265 | ClinGen Coordinates |
| 16p13.11 BP II-III | chr16:15386338-18291982 | hg18 | Ullmann\_2007[340](#_ENREF_340); Hannes\_2008[341](#_ENREF_341); de Kovel\_2010[342](#_ENREF_342); Ingason\_2011[343](#_ENREF_343) | chr16:15478837-18384481 | Coordinates correspond to Sample ‘Ao1461’ from de Kovel *et al.* |
| 16p12.2-p11.2 | chr16:21463739-29272756 | GRCh38 | Ballif\_2007[344](#_ENREF_344); Battaglia\_2009[345](#_ENREF_345); Hempel\_2009[346](#_ENREF_346);  Tabet\_2012[347](#_ENREF_347); Barber\_2013[348](#_ENREF_348) | chr16:21475060-29284077 | DECIPHER Coordinates |
| chr16:21500741-30188533 | GRCh38 | chr16:21512063-30199854 | DECIPHER Coordinates |
| 16p12.1 | chr16:21935203-22455963 | GRCh38 | DECIPHER | chr16:21946524-22467284 | DECIPHER Coordinates |
| 16p11.2 | chr16:28822635-29046499 | hg19 | Ghebranious\_2007[349](#_ENREF_349); Weiss\_2008[350](#_ENREF_350); Bachmann-Gagescu\_2010[351](#_ENREF_351); Bochukova\_2010[352](#_ENREF_352); Rosenfeld\_2010[353](#_ENREF_353); Sampson\_2010[354](#_ENREF_354); Kaminsky\_2011[40](#_ENREF_40); Tabet\_2012[347](#_ENREF_347); Rosenfeld\_2013[355](#_ENREF_355) | chr16:28822635-29046499 | Coordinates correspond to the 220kb ClinGen interval |
| chr16:29649997-30199852 | hg19 | chr16:29649997-30199852 | Coordinates correspond to the 593kb ClinGen interval |
| 16q22 | chr16:66923587-67761965 | hg19 | Abdullah\_2019[356](#_ENREF_356) | chr16:66923587-67761965 | Coordinates correspond to Patient 4 of Abdullah *et al.* |
| 16q24.1 | chr16:83671523-86296478 | hg19 | Stankiewicz\_2009[357](#_ENREF_357); Alsina Casanova\_2017[358](#_ENREF_358);  Szafranski\_2018[359](#_ENREF_359); Szafranski\_2019[360](#_ENREF_360) | chr16:83671523-86296478 | Coordinates correspond to Patient 179.3 of Szafranski *et al.* |
| chr16:84374208-85277007 | hg18 | chr16:85816707-86719506 | Coordinates correspond to Patient D3 of Stankiewicz *et al.* |
| chr16:86148250-86301591 | hg19 | chr16:86148250-86301591 | Coordinates correspond to Patient 119.3 of Szafranski *et al.* |
|  |  |  |  |  |  |
| Miller-Dieker Syndrome  (17p13.3) | chr17:1247833-2588909 | hg19 | Bi\_2009[361](#_ENREF_361); Nagamani\_2009[362](#_ENREF_362); Roos\_2009[363](#_ENREF_363); Bruno\_2010[364](#_ENREF_364); Curry\_2013[365](#_ENREF_365); Barros Fontes\_2017[366](#_ENREF_366); Emrick\_2019[367](#_ENREF_367) | chr17:1247833-2588909 | ClinGen Coordinates |
| chr17:150208-2685615 | GRCh38 | chr17:1-2588909 | DECIPHER Coordinates |
| chr17:800000-1600000 | hg18 | chr17:853250-1653250 | Coordinates correspond to Group 1 defined by Curry *et al.* |
| chr17:2000000-3000000 | hg18 | chr17:2053250-3053250 | Coordinates correspond to Group 3 defined by Curry *et al.* |
| SHFLD3  (17p13.3) | chr17:950721-1145540 | hg18 | Armour\_2011[368](#_ENREF_368); Klopocki\_2012[369](#_ENREF_369); Luk\_2014[370](#_ENREF_370); Petit\_2014[371](#_ENREF_371) | chr17:1003971-1198790 | Coordinates correspond to the critical region defined by Armour *et al.* |
| 17p13.1 | chr17:6610939-8121046 | hg19 | Shlien\_2010[372](#_ENREF_372); Carvalho\_2014[373](#_ENREF_373); Giordano\_2014[374](#_ENREF_374);  Maini\_2016[375](#_ENREF_375); Leka-Emiri\_2018[376](#_ENREF_376) | chr17:6610939-8121046 | Coordinates correspond to start of Mooneyham in Leka-Emiri *et al.*to end of Patient 2 of Shlien *et al.* |
| chr17:7429371-7972019 | hg18 | chr17:7488647-8031294 | Coordinates correspond to Patient 3 of Shlien *et al.* |
| chr17:6690620-7276764 | hg19 | chr17:6690620-7276764 | Coordinates correspond to Coutton of Leka-Emri *et al.* |
| Yuan-Harel-Lupski  (17p11.2) | chr17:12434684-21531363 | hg19 | Yuan\_2015[377](#_ENREF_377); Yuan\_2016[378](#_ENREF_378);  Fernández-Hernández\_2019[379](#_ENREF_379); Beck\_2019[380](#_ENREF_380) | chr17:12434684-21531363 | Coordinates correspond to Patient 2790 of Yuan *et al.* |
| chr17:15060682-18212609 | hg19 | chr17:15060682-18212609 | Coordinates correspond to Patient 3813 of Yuan *et al.* |
| CMT1A / HNPP  (17p11.2) | chr17:14097915-15422952 | hg19 | Zhang\_2010[381](#_ENREF_381) | chr17:14097915-15422952 | ClinGen Coordinates |
| chr17:15133094-15422954 | hg19 | chr17:15133094-15422954 | Coordinates correspond to the region between PMP22 and CMT1A-REP |
| Potocki-Lupski / Smith-Magenis (17p11.2) | chr17:16810028-20213202 | hg19 | Shaw\_2004[382](#_ENREF_382); Potocki\_2007[383](#_ENREF_383);  Zhang\_2010[384](#_ENREF_384); Beck\_2019[380](#_ENREF_380); Franciskovich\_2020[385](#_ENREF_385) | chr17:16810028-20213202 | ClinGen Coordinates |
| chr17:16796512-18088067 | hg19 | chr17:16796512-18088067 | Coordinates correspond to critical region defined by Potocki *et al.* between RP11-48J14; RP11-258F1 |
| chr17:15691101-20635656 | hg19 | chr17:15691101-20635656 | Coordinates correspond to 5Mb Interval (Shaw *et al.*): CTD-3157E16 to RP11-218E15 |
| NF1 Type I (17q11.2) | chr17:28961884-30370955 | hg19 | Kehrer‑Sawatzki\_2004[386](#_ENREF_386); Moles\_2012[387](#_ENREF_387); Serra\_2019[388](#_ENREF_388) | chr17:28961884-30370955 | Coordinates correspond to the region between the midpoints of REP-A and REP-C |
| NF1 Type II (17q11.2) | chr17:29097069-30264027 | hg19 | Kehrer‑Sawatzki\_2004[386](#_ENREF_386); Moles\_2012[387](#_ENREF_387); Serra\_2019[388](#_ENREF_388) | chr17:29097069-30264027 | ClinGen Coordinates |
| NF1 Type III (17q11.2) | chr17:29465301-30478590 | hg19 | Kehrer‑Sawatzki\_2004[386](#_ENREF_386); Moles\_2012[387](#_ENREF_387); Serra\_2019[388](#_ENREF_388) | chr17:29465301-30478590 | Coordinates correspond to the region between the start of REP-B and end of REP-C |
| RCAD Syndrome (17q12) | chr17:36459259-37856298 | GRCh38 | DECIPHER | chr17:34815073-36215917 | DECIPHER Coordinates |
| Koolen De Vries (17q21.31) | chr17:45627800-46217040 | GRCh38 | DECIPHER | chr17:43705166-44294406 | DECIPHER Coordinates |
| 17q23.1-23.2 | chr17:58113002-60275809 | hg19 | Alvarado\_2010[389](#_ENREF_389); Ballif\_2010[390](#_ENREF_390);  Schönewolf-Greulich\_2011[391](#_ENREF_391); Peterson\_2014[392](#_ENREF_392) | chr17:58113002-60275809 | ClinGen Coordinates |
| 17q24.2-q24.3 | chr17:66865466-68134760 | hg19 | Sun\_2009[393](#_ENREF_393); DeStefano\_2014[394](#_ENREF_394); Afifi\_2015[395](#_ENREF_395); Hayashi\_2017[396](#_ENREF_396) | chr17:66865466-68134760 | Coordinates correspond to the patient reported in DeStefano *et al.* |
| chr17:64641291-65042219 | hg18 | chr17:67129696-67530624 | Coordinates correspond to patient “GD” reported in Sun *et al.* |
| 46 XX,XY Sex Reversal  (17q24.3) | chr17:67892996-69792434 | hg19 | Benko\_2009[397](#_ENREF_397); Kurth\_2009[398](#_ENREF_398);  Benko\_2011[399](#_ENREF_399); Vetro\_2011[400](#_ENREF_400); Kim\_2015[401](#_ENREF_401) | chr17:67892996-69792434 | ClinGen Coordiantes |
| chr17:69477571-69510055 | hg19 | chr17:69477571-69510055 | Coordinates correspond to the 46,XY SRO defined by Kim *et al.* |
| chr17:69533161-69601161 | hg19 | chr17:69533161-69601161 | Coordinates correspond to the 46,XX SRO defined by Kim *et al.* |
|  |  |  |  |  |  |
| 18p | chr18:1-15400000 | hg19 | Wester\_2006[402](#_ENREF_402); Turleau\_2008[403](#_ENREF_403); Sebold\_2010[404](#_ENREF_404); O'Donnell\_2015[405](#_ENREF_405) | chr18:1-15400000 | Coordinates correspond to p-arm of chr18 |
| 18q Proximal | chr18:19886814-27306978 | hg19 | Cody\_2007[406](#_ENREF_406); Feenstra\_2007[407](#_ENREF_407); Heard\_2009[408](#_ENREF_408);  Cody\_2015[409](#_ENREF_409); Rojnueangnit\_2019[410](#_ENREF_410) | chr18:19886814-27306978 | Coordinates correspond to Patient 1 of Rojnueangnit *et al.* |
| chr18:28354894-41068920 | hg19 | chr18:28354894-41068920 | Coordinates correspond to DECIPHER Case 286198 referenced in Rojnueangnit *et al.* |
| chr18:41756428-44600418 | hg19 | chr18:41756428-44600418 | Coordinates correspond to DECIPHER Case 249099 referenced in Rojnueangnit *et al.* |
| 18q Terminal | chr18:52889429-78077248 | hg19 | Heard\_2009[408](#_ENREF_408); Hasi\_2014[411](#_ENREF_411); Cody\_2014[412](#_ENREF_412); Cody\_2015[409](#_ENREF_409) | chr18:52889429-78077248 | Coordinates correspond to the region between *TCF4* and 18qter |
| chr18:61637263-78077248 | hg19 | chr18:61637263-78077248 | Coordinates correspond to the Distal 18q- Reference Group |
| 19p13.3 | chr19:1952590-6908729 | hg19 | Ishikawa\_2013[413](#_ENREF_413); Nevado\_2015[414](#_ENREF_414); Orellana\_2015[415](#_ENREF_415); Novikova\_2017[416](#_ENREF_416); Tenorio\_2019[417](#_ENREF_417) | chr19:1952590-6908729 | Coordinates correspond to Patients 1-3 in Orellana *et al.* |
| chr19:3979568-4093035 | hg19 | chr19:3979568-4093035 | Coordinates correspond to the SRO defined by Nevado *et al.* |
| 19p13.2 | chr19:12793474-13104643 | hg18 | Dolan\_2010[418](#_ENREF_418); Malan\_2010[419](#_ENREF_419);  Shimojima\_2015[420](#_ENREF_420); Jezela-Stanek\_2016[421](#_ENREF_421) | chr19:12932475-13243643 | Coordinates correspond to Patient 3 of Dolan *et al.* |
| chr19:12527157-13563832 | hg19 | chr19:12527157-13563832 | Coordinates correspond to Patient 3 of Shimojima *et al.* |
| 19p13.12 | chr19:14130000-16050000 | hg18 | Jensen\_2009[422](#_ENREF_422); Bonaglia\_2010[423](#_ENREF_423); Gallant\_2011[424](#_ENREF_424);  Van der Aa\_2011[425](#_ENREF_425); de Souza\_2017[426](#_ENREF_426) | chr19:14269000-16189000 | Coordinates correspond to Patient 1 of Bonaglia *et al.* |
| 19q13.11 Proximal | chr19:33509029-37420026 | hg18 | Chowdhury\_2014[427](#_ENREF_427); Caubit\_2016[428](#_ENREF_428) | chr19:28817189-32728186 | Coordinates correspond to Chowdhury *et al* Patient 4 |
| chr19:31765851-31840190 | hg19 | chr19:31765851-31840190 | *TSHZ3* Coordinates |
| 19q13.11 Distal | chr19:37301826-41577180 | hg18 | Malan\_2009[429](#_ENREF_429); Gana\_2012[430](#_ENREF_430); Chowdhury\_2016[427](#_ENREF_427); Abe\_2018[431](#_ENREF_431) | chr19:32609986-36885340 | Coordinates correspond to Malan *et al* Patient 2 |
| chr19:39803651-40127916 | hg18 | chr19:35111811-35436076 | Coordinates correspond to SRO defined by Gana *et al.* |
| 19q13.2 | chr19:42492136-42756726 | hg19 | Yuan\_2016[432](#_ENREF_432); Kessi\_2018[433](#_ENREF_433); Calpena\_2021[434](#_ENREF_434); Singh\_2021[435](#_ENREF_435) | chr19:42492136-42756726 | Coordinates correspond to Subject 5 of Calpena *et al.* |
| chr19:42690571-42766082 | hg19 | chr19:42690571-42766082 | Coordinates correspond to Patient 3 of Singh *et al.* |
|  |  |  |  |  |  |
| 20p13 | chr20:266000-1676018 | hg19 | McGill\_2010[436](#_ENREF_436); Moutton\_2012[437](#_ENREF_437); An\_2013[438](#_ENREF_438); Fang\_2019[439](#_ENREF_439) | chr20:266000-1676018 | Coordinates correspond to Patient 1 of An *et al.* |
| chr20:102422-508612 | hg19 | chr20:102422-508612 | Coordinates correspond to Patient 6 of An *et al.* |
| chr20:297444-344879 | hg19 | chr20:297444-344879 | Coordinates correspond to the SRO defined by An *et al.* |
| 20p12.3 | chr20:6222266-9042183 | hg18 | Lalani\_2009[440](#_ENREF_440); Sahoo\_2011[441](#_ENREF_441); Williams\_2012[442](#_ENREF_442); Tan\_2017[443](#_ENREF_443) | chr20:6274266-9094183 | Coordinates correspond to the start of Patient 1 to the end of Patient 3 of Sahoo *et al.* |
| chr20:6222266-6814990 | hg18 | chr20:6274266-6866990 | Coordinates correspond to Patient 1 of Sahoo *et al.* |
| 20q13.33 | chr20:60760865-62379119 | hg18 | Traylor\_2010[444](#_ENREF_444); Mefford\_2012[445](#_ENREF_445); Mosca-Boidron\_2013[446](#_ENREF_446) | chr20:61290420-62908675 | Coordinates correspond to Patient 2 of Traylor *et al.* |
|  |  |  |  |  |  |
| 21q22 | chr21:35337577-41102161 | hg18 | Oegema\_2010[447](#_ENREF_447); Valetto\_2012[448](#_ENREF_448); Ji\_2015[449](#_ENREF_449) | chr21:36415707-42180291 | Coordinates correspond to start of Patient 2 to end of Patient 1 in Oegema *et al.* |
| chr21:37450000-37931000 | hg18 | chr21:38528131-39009130 | Coordinates correspond to Yamamoto Patient 5 from Valetto *et al.* |
| APP (21q21.3) | chr21:25880549-26171128 | GRCh38 | DECIPHER | chr21:27252860-27543446 | DECIPHER Coordinates |
|  |  |  |  |  |  |
| Cat-eye  (22q11.1-q11.21) | chr22:17392953-18591860 | hg19 | McDermid\_2002[450](#_ENREF_450); Kriek\_2006[451](#_ENREF_451); Zhang\_2015[452](#_ENREF_452) | chr22:17392953-18591860 | ClinGen Coordinates |
| chr22:17392953-20509428 | hg19 | chr22:17392953-20509428 | Coordinates correspond to the region between ClinGen CES and 22q11.2 ‘B’ breakpoint |
| chr22:17392953-24021303 | hg19 | chr22:17392953-24021303 | Coordinates correspond to the region between ClinGen CES and 22q11.2 ‘F’ breakpoint |
| DGS/VCFS AB (22q11.2) | chr22:18912231-20287208 | hg19 | Gottlieb\_1998[453](#_ENREF_453); McDermid\_2002[450](#_ENREF_450); Kaminsky\_2011[40](#_ENREF_40); Burnside\_2015[454](#_ENREF_454); Morrow\_2018[455](#_ENREF_455) | chr22:18912231-20287208 | ClinGen Coordinates |
| DGS/VCFS AC (22q11.2) | chr22:18832752-21047287 | hg19 | Burnside\_2015[454](#_ENREF_454); Morrow\_2018[455](#_ENREF_455) | chr22:18832752-21047287 | Coordinates correspond to the region between the midpoints of breakpoints A,C |
| DGS/VCFS AD (22q11.2) | chr22:18912231-21465672 | hg19 | Gottlieb\_1998[453](#_ENREF_453); McDermid\_2002[450](#_ENREF_450); Kaminsky\_2011[40](#_ENREF_40); Burnside\_2015[454](#_ENREF_454); Morrow\_2018[455](#_ENREF_455) | chr22:18912231-21465672 | ClinGen Coordinates |
| DGS/VCFS BD (22q11.2) | chr22:20287208-21465672 | hg19 | Rump\_2014; [456](#_ENREF_456); Burnside\_2015[454](#_ENREF_454); Morrow\_2018[455](#_ENREF_455) | chr22:20287208-21465672 | ClinGen Coordinates |
| DGS/VCFS CD (22q11.2) | chr22:21047287-21640885 | hg19 | Rump\_2014[456](#_ENREF_456); Burnside\_2015[454](#_ENREF_454); Woodward\_2017[457](#_ENREF_457); Morrow\_2018[455](#_ENREF_455) | chr22:21047287-21640885 | Coordinates correspond to the region between the midpoints of breakpoints C,D |
| DGS/VCFS Distal (22q11.2) | chr22:21917117-23649111 | hg19 | Rauch\_2005[458](#_ENREF_458); Tan\_2011[459](#_ENREF_459); Mikhail\_2014[460](#_ENREF_460); Burnside\_2015[454](#_ENREF_454) | chr22:21917117-23649111 | ClinGen Coordinates |
| chr22:21640885-25041769 | hg19 | chr22:21640885-25041769 | Coordinates correspond to the region between the midpoints of breakpoints D,H |
| Phelan-McDermid (22q13.33) | chr22:41673833-42839139 | hg19 | Dhar\_2009[461](#_ENREF_461); Phelan\_2011[462](#_ENREF_462); Han\_2013[463](#_ENREF_463); Disciglio\_2014[464](#_ENREF_464); Phelan\_2015[465](#_ENREF_465); Upadia\_2017[466](#_ENREF_466) | chr22:41673833-42839139 | Coordinates correspond to patient reported in Upadia *et al.* |
| chr22:43709818-50692268 | hg19 | chr22:43709818-50692268 | Coordinates correspond to Patient 8 of Disciglio *et al.* |
| chr22:46458243-49499770 | hg18 | chr22:48079579-51152904 | Coordinates correspond to Patient 5 of Dhar *et al.* |
| chr22:48554059-49565875 | hg18 | chr22:50168055-51219009 | Coordinates correspond to Patient 4 of Dhar *et al.* |
| chr22:50607088-50749416 | GRCh38 | chr22:51045516-51187844 | DECIPHER Coordinates |
|  |  |  |  |  |  |
| SHOX (Xp22.33) | chrX:791143-907140 | GRCh38 | DECIPHER | chrX:751878-867875 | DECIPHER Coordinates |
| chrX:499823-793142 | GRCh38 | chrX:460558-753877 | DECIPHER Coordinates |
| STS (Xp22.31) | chrX:6455812-8124954 | hg19 | Shapiro\_1989[467](#_ENREF_467); Alperin\_1997[468](#_ENREF_468); Valdes-Flores\_2000[469](#_ENREF_469);  Hand\_2014[470](#_ENREF_470); Brcic\_2020[471](#_ENREF_471); Gubb\_2020[472](#_ENREF_472) | chrX:6455812-8124954 | ClinGen Coordinates |
| chrX:7065293-7272684 | hg19 | chrX:7065293-7272684 | ClinGen Coordinates |
| Xp21 | chrX:28605681-33229673 | hg19 | Hellerud\_2003[473](#_ENREF_473); Stanczak\_2007[474](#_ENREF_474); Heide\_2015[475](#_ENREF_475); Korkut\_2016[476](#_ENREF_476); Montoya-Williams\_2017[477](#_ENREF_477) | chrX:28605681-33229673 | Coordinates correspond to the region between *IL1RAPL1* and *DMD* |
| Xp11.3 | chrX:46336830-46740394 | hg19 | Lugtenberg\_2006[478](#_ENREF_478); Zhang\_2006[479](#_ENREF_479); Delphin\_2012[480](#_ENREF_480) | chrX:46336830-46740394 | Coordinates correspond to Family 2 of Delphin *et al.* |
| Xp11.23-p11.22 | chrX:48306152-52103258 | hg19 | Giroda\_2009[481](#_ENREF_481); Nizon\_2015[482](#_ENREF_482); Grams\_2016[483](#_ENREF_483) | chrX:48306152-52103258 | ClinGen Coordinates |
| Xp11.22 | chrX:53363456-53793054 | hg19 | Whibley\_2010[484](#_ENREF_484); Froyen\_2012[485](#_ENREF_485); Grams\_2016[483](#_ENREF_483) | chrX:53363456-53793054 | ClinGen Coordinates |
| Xq21 | chrX:80817978-88868979 | hg19 | Song\_2010[486](#_ENREF_486); Iossa\_2015[487](#_ENREF_487); Liang\_2017[488](#_ENREF_488) | chrX:80817978-88868979 | Coordinates correspond to the patient reported in Liang *et al.* |
| chrX:82763269-85302566 | hg19 | chrX:82763269-85302566 | Coordinates correspond to the region between *POU3F4* and *CHM* |
| Pelizaeus-Merzbacher (Xq22.2) | chrX:103776510-103792618 | GRCh38 | DECIPHER | chrX:103031438-103047547 | DECIPHER Coordinates |
| DL-ATS (Xq22.3) | chrX:107398837-107940775 | hg19 | Meloni\_2002[489](#_ENREF_489); Uliana\_2011[490](#_ENREF_490); Nozu\_2017[491](#_ENREF_491) | chrX:107398837-107940775 | Coordinates correspond to the region between *COL4A6* and *COL4A5* |
| MR-ATS (Xq22.3) | chrX:107683074-108976621 | hg19 | Meloni\_2002[489](#_ENREF_489); Hoischen\_2009[492](#_ENREF_492); Rodriguez\_2010[493](#_ENREF_493) | chrX:107683074-108976621 | Coordinates correspond to the region between *COL4A5* and *ACSL4* |
| Xq25 | chrX:122673975-123546561 | hg19 | Di Benedetto\_2014[494](#_ENREF_494); Yingjun\_2015[495](#_ENREF_495); Leroy\_2016[496](#_ENREF_496); Turchi\_2020[497](#_ENREF_497) | chrX:122673975-123546561 | Coordinates correspond to Patient reported in Turchi *et al.* |
| XLAG (Xq26.3) | chrX:135627637-136118269 | hg19 | Trivellin­\_2014[498](#_ENREF_498); Iacovazzo\_2016[499](#_ENREF_499); Naves\_2016[500](#_ENREF_500) | chrX:135627637-136118269 | Coordinates correspond to the region between SRO1 and SRO2 defined by Trivellin *et al.* |
| Xq27.1 | chrX:139532854-140080654 | hg19 | Sutton\_2011[501](#_ENREF_501); Moalem\_2012[502](#_ENREF_502); Vetro\_2015[503](#_ENREF_503); Tasic\_2019[504](#_ENREF_504) | chrX:139532854-140080654 | Coordinates correspond to patient reported in Tasic *et al.* |
| chrX:139178566-139675085 | hg19 | Bowl\_2005[505](#_ENREF_505); Arya\_2019[506](#_ENREF_506); Rosolowsky\_2020[507](#_ENREF_507) | chrX:139178566-139675085 | Coordinates correspond to patient reported in Rosolowsky *et al.* |
| Xq27.3-q28 | chrX:146789031-147152262 | hg19 | Rio\_2009[508](#_ENREF_508); Nagamani\_2012[509](#_ENREF_509); Vengoechea\_2012[510](#_ENREF_510); Hickey\_2013[511](#_ENREF_511) | chrX:146789031-147152262 | Coordinates correspond to Patient 1 of Nagamani *et al.* |
| chrX:146993469-147032647 | hg19 | chrX:146993469-147032647 | *FMR1* Coordinates |
| MECP2 (Xq28) | chrX:153287264-153363188 | hg19 | Amir\_1999[512](#_ENREF_512); Van\_Esch\_2012[513](#_ENREF_513);  Vidal\_2019[514](#_ENREF_514); Pascual-Alonso\_2020[515](#_ENREF_515) | chrX:153287264-153363188 | ClinGen Coordinates |
| chrX:152957295-153594098 | hg19 | chrX:152957295-153594098 | Coordinates correspond to Patient 14 of Pascual-Alonso *et al*. |
| Xq28 (GDI1) | chrX:154396222-154653579 | GRCh38 | Froyen\_2007[516](#_ENREF_516); Madrigal\_2007[517](#_ENREF_517);  Vandewalle\_2009[518](#_ENREF_518); Sinibaldi\_2019[519](#_ENREF_519) | chrX:153624563-153881853 | DECIPHER Coordinates |
| Xq28 (RAB39B) | chrX:154118603-154564401 | hg19 | El-Hattab\_2011[520](#_ENREF_520); Vanmarsenille\_2014[521](#_ENREF_521); El-Hattab\_2015[522](#_ENREF_522) | chrX:154118603-154564401 | ClinGen Coordinates |
| Xq28 Moyamoya | chrX:154210613-154364362 | hg19 | Hervé\_2010[523](#_ENREF_523); Miskinyte\_2011[524](#_ENREF_524); Janczar\_2016[525](#_ENREF_525); Jourdy\_2017[526](#_ENREF_526) | chrX:154210613-154364362 | Coordinates correspond to patient reported in Janczar *et al.* |
| chrX:154299695-154351349 | hg19 | chrX:154299695-154351349 | *BRCC3* Coordinates |
|  |  |  |  |  |  |
| AZFa (Yq11.21) | chrY:12232056-13042949 | GRCh38 | DECIPHER | chrY:14352761-15154862 | DECIPHER Coordinates |
| AZFb (Yq11.222-q11.223) | chrY:18006165-23919050 | GRCh38 | DECIPHER | chrY:20118045-26065197 | DECIPHER Coordinates |
| AZFb+AZFc (Yq11.222-q11.23) | chrY:17852946-25647683 | GRCh38 | DECIPHER | chrY:19964826-27793830 | DECIPHER Coordinates |
| AZFc (Yq11.223-q11.23) | chrY:22831278-25887782 | GRCh38 | DECIPHER | chrY:24977425-28033929 | DECIPHER Coordinates |

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