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| **Genetic Disorder** | **Description** | **Reference** | **RNA Variant** | **Amino acid substitution** |
| Achondroplasia | Achondroplasia is a genetic disorder whose primary feature is dwarfism. In those with the condition, the arms and legs are short, while the torso is typically of normal length. | Pauli, RM; Adam, MP; Ardinger, HH; Pagon, RA; Wallace, SE; Bean, LJH; Mefford, HC; Stephens, K; Amemiya, A; Ledbetter, N (2012). "Achondroplasia". GeneReviews. PMID 20301331 | NM\_000142.5(FGFR3):c.1123G>T | NP\_000133.1:p.Gly375Cys |
| Huntington's disease | Huntington's disease (HD), also known as Huntington's chorea, is a neurodegenerative disease that is mostly inherited. The earliest symptoms are often subtle problems with mood or mental abilities. A general lack of coordination and an unsteady gait often follow. | Dayalu P, Albin RL (February 2015). "Huntington disease: pathogenesis and treatment". Neurologic Clinics. 33 (1): 101–14. doi:10.1016/j.ncl.2014.09.003. PMID 25432725. | NM\_002111.8(HTT):c.93\_114del | NP\_002102.4:p.Gln31fs |
| Marfan syndrome | Marfan syndrome (MFS) is a genetic disorder that affects the connective tissue. Those with the condition tend to be tall and thin, with long arms, legs, fingers, and toes | Siepe, M; Löffelbein, F (2009). "[The Marfan syndrome and related connective tissue disorders]". Medizinische Monatsschrift für Pharmazeuten. 32 (6): 213–9. PMID 19554831. | NM\_003036.4(SKI):c.539C>A | NP\_003027.1:p.Thr180Lys |
| Polycystic kidney disease 1 | Polycystic kidney disease is a genetic disorder in which the renal tubules become structurally abnormal, resulting in the development and growth of multiple cysts within the kidney. | "Polycystic kidney disease: the complete structure of the PKD1 gene and its protein. The International Polycystic Kidney Disease Consortium". Cell. 81 (2): 289–98. April 1995. doi:10.1016/0092-8674(95)90339-9. PMID 7736581. S2CID 11114706. | NM\_001408.3(CELSR2):c.8235\_8246dup | NP\_001399.1:p.Glu2749\_Glu2752dup |
| Polycystic kidney disease 2 | Polycystic kidney disease is a genetic disorder in which the renal tubules become structurally abnormal, resulting in the development and growth of multiple cysts within the kidney. | Li, Qiang; Shen Patrick Y; Wu Guanqing; Chen Xing-Zhen (January 2003). "Polycystin-2 interacts with troponin I, an angiogenesis inhibitor". Biochemistry. United States. 42 (2): 450–7. doi:10.1021/bi0267792. ISSN 0006-2960. PMID 12525172 | NM\_000297.4(PKD2):c.136\_210delinsGCGGG | NP\_000288.1:p.Leu46fs |
| Familial hypercholesterolemia | Familial hypercholesterolemia (FH) is a genetic disorder characterized by high cholesterol levels, specifically very high levels of low-density lipoprotein (LDL) in the blood and early cardiovascular disease. | Goldberg, AC; Hopkins, PN; Toth, PP; Ballantyne, CM; Rader, DJ; Robinson, JG; Daniels, SR; Gidding, SS; de Ferranti, SD; Ito, MK; McGowan, MP; Moriarty, PM; Cromwell, WC; Ross, JL; Ziajka, PE; National Lipid Association Expert Panel on Familial, Hypercholesterolemia. (June 2011). "Familial hypercholesterolemia: screening, diagnosis and management of pediatric and adult patients: clinical guidance from the National Lipid Association Expert Panel on Familial Hypercholesterolemia". Journal of Clinical Lipidology. 5 (3 Suppl): S1–8. doi:10.1016/j.jacl.2011.04.003. PMID 21600525. | NM\_015627.3(LDLRAP1):c.71dup | NP\_056442.2:p.Gly25fs |
| Sickle cell anaemia | Sickle cell disease (SCD) is a group of blood disorders typically inherited from a person's parents. The most common type is known as sickle cell anaemia (SCA). | Yawn BP, Buchanan GR, Afenyi-Annan AN, Ballas SK, Hassell KL, James AH, Jordan L, Lanzkron SM, Lottenberg R, Savage WJ, Tanabe PJ, Ware RE, Murad MH, Goldsmith JC, Ortiz E, Fulwood R, Horton A, John-Sowah J (September 2014). "Management of sickle cell disease: summary of the 2014 evidence-based report by expert panel members". JAMA. 312 (10): 1033–48. doi:10.1001/jama.2014.10517. PMID 25203083. S2CID 37681044 | NM\_000518.5(HBB):c.79G>A | NP\_000509.1:p.Glu27Lys |
| Cystic fibrosis | Cystic fibrosis (CF) is a genetic disorder that affects mostly the lungs, but also the pancreas, liver, kidneys, and intestine. | Allen JL, Panitch HB, Rubenstein RC (2016). Cystic Fibrosis. CRC Press. p. 92. ISBN 9781439801826. Archived from the original on 2017-09-08. | NM\_015102.5(NPHP4):c.4115T>C | NP\_055917.1:p.Leu1372Pro |
| Tay–Sachs disease | Tay–Sachs disease is a genetic disorder that results in the destruction of nerve cells in the brain and spinal cord. | "Tay–Sachs disease Information Page". National Institute of Neurological Disorders and Stroke. 14 February 2007. Archived from the original on 27 November 2011. Retrieved 10 May 2007. | NM\_000405.5(GM2A):c.160G>T | NP\_000396.2:p.Glu54Ter |
| Phenylketonuria | Phenylketonuria (PKU) is an inborn error of metabolism that results in decreased metabolism of the amino acid phenylalanine. | Al Hafid N, Christodoulou J (October 2015). "Phenylketonuria: a review of current and future treatments". Translational Pediatrics. 4 (4): 304–17. doi:10.3978/j.issn.2224-4336.2015.10.07. PMC 4728993. PMID 26835392. | NM\_000320.3(QDPR):c.661C>T | NP\_000311.2:p.Arg221Ter |
| Galactosemia | Galactosemia is a rare genetic metabolic disorder that affects an individual's ability to metabolize the sugar galactose properly. | Fensom AH, Benson PF, Blunt S (November 1974). "Prenatal diagnosis of galactosaemia". Br Med J. 4 (5941): 386–7. doi:10.1136/bmj.4.5941.386. PMC 1612460. PMID 4154122. | NM\_001008216.2(GALE):c.937C>A | NP\_001008217.1:p.Leu313Met |
| Haemophilia | Haemophilia is a mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding. | "What Is Hemophilia?". NHLBI. July 13, 2013. Archived from the original on 4 October 2016. Retrieved 8 September 2016. | NM\_000133.3(F9):c.141T>A | NP\_000124.1:p.Tyr47Ter |
| Duchenne muscular dystrophy | Duchenne muscular dystrophy (DMD) is a severe type of muscular dystrophy that primarily affects boys.Muscle weakness usually begins around the age of four, and worsens quickly | Rowland, L.P. (1985). "Clinical Perspective: Phenotypic Expression In Muscular Dystrophy". In Strohman, C.; Wolf, S. (eds.). Gene Expression in Muscle. Advances in Experimental Medicine and Biology. Plenum Press. pp. 3–5. ISBN 978-1-4684-4907-5. | NM\_000337.5(SGCD):c.89G>A | NP\_000328.2:p.Trp30Ter |
| hereditary multiple exostoses | Hereditary multiple osteochondromas (HMO), also known as hereditary multiple exostoses, is a disorder characterized by the development of multiple benign osteocartilaginous masses (exostoses) in relation to the ends of long bones of the lower limbs such as the femurs and tibias and of the upper limbs such as the humeri and forearm bones. They are also known as osteochondromas | Wuyts, W; Schmale, GA; Chansky, HA; et al. (21 November 2013). "Hereditary Multiple Osteochondromas". GeneReviews. University of Washington, Seattle. Retrieved 24 March 2018 | NM\_000127.2(EXT1):c.2132G>A | NP\_000118.2:p.Trp711Ter |
| tuberous sclerosis | Tuberous sclerosis complex (TSC) is a rare multisystem autosomal dominant genetic disease that causes non-cancerous tumours to grow in the brain and on other vital organs such as the kidneys, heart, liver, eyes, lungs and skin. | James PD, Notley C, Hegadorn C, Leggo J, Tuttle A, Tinlin S, Brown C, Andrews C, Labelle A, Chirinian Y, O'Brien L, Othman M, Rivard G, Rapson D, Hough C, Lillicrap D (January 2007). "The mutational spectrum of type 1 von Willebrand disease: Results from a Canadian cohort study". Blood. 109 (1): 145–54. doi:10.1182/blood-2006-05-021105. PMID 17190853. | NM\_000488.3(SERPINC1):c.1213C>A | NP\_000479.1:p.Leu405Ile |
| Von Willebrand disease | Von Willebrand disease (VWD) is the most common hereditary blood-clotting disorder in humans. An acquired form can sometimes result from other medical conditions. It arises from a deficiency in the quality or quantity of von Willebrand factor (VWF), a multimeric protein that is required for platelet adhesion | James PD, Notley C, Hegadorn C, Leggo J, Tuttle A, Tinlin S, Brown C, Andrews C, Labelle A, Chirinian Y, O'Brien L, Othman M, Rivard G, Rapson D, Hough C, Lillicrap D (January 2007). "The mutational spectrum of type 1 von Willebrand disease: Results from a Canadian cohort study". Blood. 109 (1): 145–54. doi:10.1182/blood-2006-05-021105. PMID 17190853. |  |  |
| Albinism, oculocutaneous, type II | Oculocutaneous albinism is a form of albinism involving the eyes (oculo-), the skin (-cutaneous), and the hair. | "Oculocutaneous albinism". Genetics Home Reference. U.S. National Library of Medicine. Retrieved 5 August 2020. | NM\_000550.3(TYRP1):c.497C>G | NP\_000541.1:p.Ser166Ter |
| Niemann–Pick disease | Niemann–Pick disease is a group of severe inherited metabolic disorders, in which sphingomyelin accumulates in lysosomes in cells | "Neimann-Pick Disease". Genetics Home Reference. NIH. January 2008. Retrieved 2 October 2012. | NM\_000543.5(SMPD1):c.28C>T | NP\_000534.3:p.Gln10Ter |
| spinal muscular atrophy | Spinal muscular atrophy (SMA) is a rare neuromuscular disorder that results in the loss of motor neurons and progressive muscle wasting. | Prior, Thomas W.; Leach, Meganne E.; Finanger, Erika (1993), Adam, Margaret P.; Ardinger, Holly H.; Pagon, Roberta A.; Wallace, Stephanie E. (eds.), "Spinal Muscular Atrophy", GeneReviews®, Seattle (WA): University of Washington, Seattle, PMID 20301526, retrieved 25 October 2020 | NM\_020631.5(PLEKHG5):c.2542C>T | NP\_001036128.1:p.Arg904Ter |
| Roberts syndrome | Roberts syndrome, syndrome, is an extremely rare autosomal recessive genetic disorder that is characterized by mild to severe prenatal retardation or disruption of cell division, leading to malformation of the bones in the skull, face, arms, and legs. | Kugler, Mary. "Roberts syndrome: Inherited Disorder Causes Abnormal Bone Development." About.com: Rare Diseases. About. 23 April 2005. | NM\_000975.5(RPL11):c.204del | NP\_000966.2:p.Ile68fs |
| Rett syndrome | Rett syndrome (RTT) is a genetic disorder that typically becomes apparent after 6–18 months of age in female. | "Rett Syndrome". NORD (National Organization for Rare Disorders). 2015. Archived from the original on 19 February 2017. Retrieved 14 October 2017. | NM\_005458.8(GABBR2):c.1699G>A | NP\_005449.5:p.Ala567Thr |
| Down syndrome | Down syndrome or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. | Weijerman, ME; de Winter, JP (Dec 2010). "Clinical practice. The care of children with Down syndrome". European Journal of Pediatrics. 169 (12): 1445–52. doi:10.1007/s00431-010-1253-0. PMC 2962780. PMID 20632187. | NM\_198576.4(AGRN):c.5179G>T | NP\_940978.2:p.Val1727Phe |
| Turner syndrome | Turner syndrome (TS), also known 45,X, or 45,X0, is a genetic condition in which a female is partly or completely missing an X chromosome | Turner Syndrome: Overview". Eunice Kennedy Shriver National Institute of Child Health and Human Development. 3 April 2013. Archived from the original on 2 April 2015. Retrieved 15 March 2015. | NM\_153818.1(PEX10):c.764dup | NP\_002608.1:p.Leu236fs |
| Aicardi syndrome | Aicardi syndrome is a rare genetic malformation syndrome characterized by the partial or complete absence of a key structure in the brain called the corpus callosum, the presence of retinal abnormalities, and seizures in the form of infantile spasms. | Sijmons, Rolf H (2008). "Encyclopaedia of tumour-associated familial disorders. Part I: from AIMAH to CHIME syndrome". Hereditary Cancer in Clinical Practice. 6 (1): 22–57. doi:10.1186/1897-4287-6-1-22. PMC 2735164. PMID 19706204. | NM\_001111.5(ADAR):c.3363dup | NP\_001102.3:p.Lys1122Ter |
| Edward syndrome | Edwards syndrome, also known as trisomy 18, is a genetic disorder caused by the presence of a third copy of all or part of chromosome 18. | "Edwards syndrome (John Hilton Edwards)". WhoNamedIt.com. Archived from the original on 2008-07-09. Retrieved 2008-07-24. | NM\_000465.4(BARD1):c.2268G>A | NP\_000456.2:p.Trp756Ter |