6 References

1. Boycott KM, Vanstone MR, Bulman DE, MacKenzie AE. Rare-disease genetics in the era of next-generation sequencing: discovery to translation. Nature reviews Genetics. 2013 Oct;14(10):681-91. PubMed PMID: 23999272.

2. Rilstone JJ, Alkhater RA, Minassian BA. Brain dopamine-serotonin vesicular transport disease and its treatment. N Engl J Med. 2013 Feb 7;368(6):543-50. PubMed PMID: 23363473.

3. Beaulieu CL, Majewski J, Schwartzentruber J, Samuels ME, Fernandez BA, Bernier FP, et al. FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. Am J Hum Genet. 2014 Jun 5;94(6):809-17. PubMed PMID: 24906018.

4. Wu CH, Fallini C, Ticozzi N, Keagle PJ, Sapp PC, Piotrowska K, et al. Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature. 2012 Aug 23;488(7412):499-503. PubMed PMID: 22801503. Pubmed Central PMCID: 3575525.

5. Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, et al. Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nat Genet. 2012 Apr;44(4):376-8. PubMed PMID: 22426308.

6. Tsui LC, Buchwald M, Barker D, Braman JC, Knowlton R, Schumm JW, et al. Cystic fibrosis locus defined by a genetically linked polymorphic DNA marker. Science. 1985 Nov 29;230(4729):1054-7. PubMed PMID: 2997931.

7. Ng SB, Buckingham KJ, Lee C, Bigham AW, Tabor HK, Dent KM, et al. Exome sequencing identifies the cause of a mendelian disorder. Nat Genet. 2010 Jan;42(1):30-5. PubMed PMID: 19915526. Pubmed Central PMCID: 2847889.

8. Wang Z, Liu X, Yang BZ, Gelernter J. The role and challenges of exome sequencing in studies of human diseases. Frontiers in genetics. 2013;4:160. PubMed PMID: 24032039. Pubmed Central PMCID: 3752524.

9. Firth HV, Wright CF. The Deciphering Developmental Disorders (DDD) study. Dev Med Child Neurol. 2011 Aug;53(8):702-3. PubMed PMID: 21679367. Epub 2011/06/18. eng.

10. Kaye J, Hurles M, Griffin H, Grewal J, Bobrow M, Timpson N, et al. Managing clinically significant findings in research: the UK10K example. Eur J Hum Genet. 2014 Jan 15. PubMed PMID: 24424120. Pubmed Central PMCID: 4026295.

11. Yang Y, Muzny DM, Reid JG, Bainbridge MN, Willis A, Ward PA, et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. N Engl J Med. 2013 Oct 17;369(16):1502-11. PubMed PMID: 24088041.

12. MacArthur DG, Manolio TA, Dimmock DP, Rehm HL, Shendure J, Abecasis GR, et al. Guidelines for investigating causality of sequence variants in human disease. Nature. 2014 Apr 24;508(7497):469-76. PubMed PMID: 24759409.

13. Tarpey PS, Smith R, Pleasance E, Whibley A, Edkins S, Hardy C, et al. A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nat Genet. 2009 May;41(5):535-43. PubMed PMID: 19377476. Pubmed Central PMCID: 2872007. Epub 2009/04/21. eng.

14. Purcell SM, Moran JL, Fromer M, Ruderfer D, Solovieff N, Roussos P, et al. A polygenic burden of rare disruptive mutations in schizophrenia. Nature. 2014 Feb 13;506(7487):185-90. PubMed PMID: 24463508.

15. Bhattacharya S, Das A, Ghosh S, Dasgupta R, Bagchi A. Hypoglycosylation of dystroglycan due to T192M mutation: a molecular insight behind the fact. Gene. 2014 Mar 1;537(1):108-14. PubMed PMID: 24361964.

16. Buysse K, Riemersma M, Powell G, van Reeuwijk J, Chitayat D, Roscioli T, et al. Missense mutations in beta-1,3-N-acetylglucosaminyltransferase 1 (B3GNT1) cause Walker-Warburg syndrome. Hum Mol Genet. 2013 Jan 28. PubMed PMID: 23359570. Epub 2013/01/30. Eng.

17. Thornhill P, Bassett D, Lochmuller H, Bushby K, Straub V. Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of fukutin-related protein (FKRP). Brain : a journal of neurology. 2008 Jun;131(Pt 6):1551-61. PubMed PMID: 18477595. Epub 2008/05/15. eng.

18. Springett A, Morris JK. Congenital Anomaly Statistics. England and Wales. London: British Isles Network of Congenital Anomaly Registers. 2010.

19. Vlastos IM, Koudoumnakis E, Houlakis M, Nasika M, Griva M, Stylogianni E. Cleft lip and palate treatment of 530 children over a decade in a single centre. Int J Pediatr Otorhinolaryngol. 2009 Jul;73(7):993-7. PubMed PMID: 19443049.

20. Verity C, Firth H, ffrench-Constant C. Congenital abnormalities of the central nervous system. J Neurol Neurosurg Psychiatry. 2003 Mar;74 Suppl 1:i3-8. PubMed PMID: 12611928. Pubmed Central PMCID: 1765611.

21. Cereda A, Carey JC. The trisomy 18 syndrome. Orphanet J Rare Dis. 2012;7:81. PubMed PMID: 23088440. Pubmed Central PMCID: 3520824.

22. Hillman SC, McMullan DJ, Hall G, Togneri FS, James N, Maher EJ, et al. Use of prenatal chromosomal microarray: prospective cohort study and systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2013 Jun;41(6):610-20. PubMed PMID: 23512800. Epub 2013/03/21. eng.

23. Rousseau F, el Ghouzzi V, Delezoide AL, Legeai-Mallet L, Le Merrer M, Munnich A, et al. Missense FGFR3 mutations create cysteine residues in thanatophoric dwarfism type I (TD1). Hum Mol Genet. 1996 Apr;5(4):509-12. PubMed PMID: 8845844. Epub 1996/04/01. eng.

24. Tabor A, Vestergaard CH, Lidegaard O. Fetal loss rate after chorionic villus sampling and amniocentesis: an 11-year national registry study. Ultrasound Obstet Gynecol. 2009 Jul;34(1):19-24. PubMed PMID: 19504504.

25. Lo YM, Corbetta N, Chamberlain PF, Rai V, Sargent IL, Redman CW, et al. Presence of fetal DNA in maternal plasma and serum. Lancet. 1997 Aug 16;350(9076):485-7. PubMed PMID: 9274585.

26. Caspersson T, Farber S, Foley GE, Kudynowski J, Modest EJ, Simonsson E, et al. Chemical differentiation along metaphase chromosomes. Exp Cell Res. 1968 Jan;49(1):219-22. PubMed PMID: 5640698.

27. Shaffer LG, Bejjani BA. A cytogeneticist's perspective on genomic microarrays. Hum Reprod Update. 2004 May-Jun;10(3):221-6. PubMed PMID: 15140869.

28. Bauman JG, Wiegant J, Borst P, van Duijn P. A new method for fluorescence microscopical localization of specific DNA sequences by in situ hybridization of fluorochromelabelled RNA. Exp Cell Res. 1980 Aug;128(2):485-90. PubMed PMID: 6157553.

29. Schrock E, du Manoir S, Veldman T, Schoell B, Wienberg J, Ferguson-Smith MA, et al. Multicolor spectral karyotyping of human chromosomes. Science. 1996 Jul 26;273(5274):494-7. PubMed PMID: 8662537.

30. Evans MI, Henry GP, Miller WA, Bui TH, Snidjers RJ, Wapner RJ, et al. International, collaborative assessment of 146,000 prenatal karyotypes: expected limitations if only chromosome-specific probes and fluorescent in-situ hybridization are used. Hum Reprod. 1999 May;14(5):1213-6. PubMed PMID: 10325263.

31. Nickerson E, Greenberg F, Keating MT, McCaskill C, Shaffer LG. Deletions of the elastin gene at 7q11.23 occur in approximately 90% of patients with Williams syndrome. Am J Hum Genet. 1995 May;56(5):1156-61. PubMed PMID: 7726172. Pubmed Central PMCID: 1801441.

32. Ligon AH, Kashork CD, Richards CS, Shaffer LG. Identification of female carriers for Duchenne and Becker muscular dystrophies using a FISH-based approach. Eur J Hum Genet. 2000 Apr;8(4):293-8. PubMed PMID: 10854113.

33. Brackley KJ, Kilby MD, Morton J, Whittle MJ, Knight SJ, Flint J. A case of recurrent congenital fetal anomalies associated with a familial subtelomeric translocation. Prenat Diagn. 1999 Jun;19(6):570-4. PubMed PMID: 10416976.

34. Wapner RJ, Martin CL, Levy B, Ballif BC, Eng CM, Zachary JM, et al. Chromosomal microarray versus karyotyping for prenatal diagnosis. N Engl J Med. 2012 Dec 6;367(23):2175-84. PubMed PMID: 23215555. Epub 2012/12/12. eng.

35. Hillman SC, Pretlove S, Coomarasamy A, McMullan DJ, Davison EV, Maher ER, et al. Additional information from array comparative genomic hybridization technology over conventional karyotyping in prenatal diagnosis: a systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2011 Jan;37(1):6-14. PubMed PMID: 20658510.

36. Srebniak MI, Boter M, Oudesluijs GO, Cohen-Overbeek T, Govaerts LC, Diderich KE, et al. Genomic SNP array as a gold standard for prenatal diagnosis of foetal ultrasound abnormalities. Molecular cytogenetics. 2012;5(1):14. PubMed PMID: 22413963. Pubmed Central PMCID: 3328283.

37. Vanakker O, Vilain C, Janssens K, Van der Aa N, Smits G, Bandelier C, et al. Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. Eur J Med Genet. 2014 Feb 15. PubMed PMID: 24534801.

38. Hillman SC, McMullan DJ, Maher ER, Kilby MD. The use of chromosomal microarray in prenatal diagnosis. The Obstetrician and Gynaecologist. 2013;15(2):80-4.

39. UKGTN. UK Genetic Testing Network arrayCGH Commissioning Workshop. London: phg foundation: 2009.

40. Valduga M, Philippe C, Bach Segura P, Thiebaugeorges O, Miton A, Beri M, et al. A retrospective study by oligonucleotide array-CGH analysis in 50 fetuses with multiple malformations. Prenat Diagn. 2010 Apr;30(4):333-41. PubMed PMID: 20155755. Epub 2010/02/16. eng.

41. Conrad DF, Keebler JE, DePristo MA, Lindsay SJ, Zhang Y, Casals F, et al. Variation in genome-wide mutation rates within and between human families. Nat Genet. 2011 Jul;43(7):712-4. PubMed PMID: 21666693. Pubmed Central PMCID: 3322360. Epub 2011/06/15. eng.

42. Mann K, Ogilvie CM. QF-PCR: application, overview and review of the literature. Prenat Diagn. 2012 Apr;32(4):309-14. PubMed PMID: 22467160.

43. Lo YM, Tein MS, Lau TK, Haines CJ, Leung TN, Poon PM, et al. Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. Am J Hum Genet. 1998 Apr;62(4):768-75. PubMed PMID: 9529358. Pubmed Central PMCID: 1377040.

44. Lun FM, Chiu RW, Allen Chan KC, Yeung Leung T, Kin Lau T, Dennis Lo YM. Microfluidics digital PCR reveals a higher than expected fraction of fetal DNA in maternal plasma. Clin Chem. 2008 Oct;54(10):1664-72. PubMed PMID: 18703764.

45. Palomaki GE, Deciu C, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, et al. DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. Genet Med. 2012 Mar;14(3):296-305. PubMed PMID: 22281937. Pubmed Central PMCID: 3938175.

46. Fan HC, Blumenfeld YJ, Chitkara U, Hudgins L, Quake SR. Analysis of the size distributions of fetal and maternal cell-free DNA by paired-end sequencing. Clin Chem. 2010 Aug;56(8):1279-86. PubMed PMID: 20558635.

47. Nicolaides KH, Syngelaki A, Gil M, Atanasova V, Markova D. Validation of targeted sequencing of single-nucleotide polymorphisms for non-invasive prenatal

detection of aneuploidy of chromosomes 13, 18, 21, X, and Y. Prenat Diagn. 2013 Jun;33(6):575-9. PubMed PMID: 23613152.

48. Ge H, Huang X, Li X, Chen S, Zheng J, Jiang H, et al. Noninvasive prenatal detection for pathogenic CNVs: the application in alpha-thalassemia. PLoS One. 2013;8(6):e67464. PubMed PMID: 23840709. Pubmed Central PMCID: 3696090.

49. Lim JH, Kim MJ, Kim SY, Kim HO, Song MJ, Kim MH, et al. Non-invasive prenatal detection of achondroplasia using circulating fetal DNA in maternal plasma. J Assist Reprod Genet. 2011 Feb;28(2):167-72. PubMed PMID: 20963478. Pubmed Central PMCID: 3059531.

50. Agarwal A, Sayres LC, Cho MK, Cook-Deegan R, Chandrasekharan S. Commercial landscape of noninvasive prenatal testing in the United States. Prenat Diagn. 2013 Jun;33(6):521-31. PubMed PMID: 23686656. Pubmed Central PMCID: 3898859.

51. Song Y, Liu C, Qi H, Zhang Y, Bian X, Liu J. Noninvasive prenatal testing of fetal aneuploidies by massively parallel sequencing in a prospective Chinese population. Prenat Diagn. 2013 Jul;33(7):700-6. PubMed PMID: 23703459.

52. Hill M, Karunaratna M, Lewis C, Forya F, Chitty L. Views and preferences for the implementation of non-invasive prenatal diagnosis for single gene disorders from health professionals in the United Kingdom. Am J Med Genet A. 2013 Jul;161A(7):1612-8. PubMed PMID: 23696422.

53. Fan HC, Gu W, Wang J, Blumenfeld YJ, El-Sayed YY, Quake SR. Non-invasive prenatal measurement of the fetal genome. Nature. 2012 Jul 19;487(7407):320-4. PubMed PMID: 22763444. Pubmed Central PMCID: 3561905.

54. Kitzman JO, Snyder MW, Ventura M, Lewis AP, Qiu R, Simmons LE, et al. Noninvasive whole-genome sequencing of a human fetus. Sci Transl Med. 2012 Jun 6;4(137):137ra76. PubMed PMID: 22674554. Pubmed Central PMCID: 3379884.

55. Veltman JA, Brunner HG. De novo mutations in human genetic disease. Nature reviews Genetics. 2012;13(8):565-75. PubMed PMID: 22805709. Epub 2012/07/19. eng.

56. O'Roak BJ, Deriziotis P, Lee C, Vives L, Schwartz JJ, Girirajan S, et al. Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nat Genet. 2011 Jun;43(6):585-9. PubMed PMID: 21572417. eng.

57. de Ligt J, Willemsen MH, van Bon BW, Kleefstra T, Yntema HG, Kroes T, et al. Diagnostic exome sequencing in persons with severe intellectual disability. N Engl J Med. 2012 Nov 15;367(20):1921-9. PubMed PMID: 23033978. Epub 2012/10/05. eng.

58. Zaidi S, Choi M, Wakimoto H, Ma L, Jiang J, Overton JD, et al. De novo mutations in histone-modifying genes in congenital heart disease. Nature. 2013 Jun 13;498(7453):220-3. PubMed PMID: 23665959. Pubmed Central PMCID: 3706629. Eng.

59. Ng SB, Turner EH, Robertson PD, Flygare SD, Bigham AW, Lee C, et al. Targeted capture and massively parallel sequencing of 12 human exomes. Nature. 2009 Sep 10;461(7261):272-6. PubMed PMID: 19684571. Pubmed Central PMCID: 2844771.

60. Glockle N, Kohl S, Mohr J, Scheurenbrand T, Sprecher A, Weisschuh N, et al. Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. Eur J Hum Genet. 2014 Jan;22(1):99-104. PubMed PMID: 23591405. Pubmed Central PMCID: 3865404.

61. Gahl WA, Markello TC, Toro C, Fajardo KF, Sincan M, Gill F, et al. The National Institutes of Health Undiagnosed Diseases Program: insights into rare diseases. Genet Med. 2012 Jan;14(1):51-9. PubMed PMID: 22237431.

62. Gilissen C, Hehir-Kwa JY, Thung DT, van de Vorst M, van Bon BW, Willemsen MH, et al. Genome sequencing identifies major causes of severe intellectual disability. Nature. 2014 Jun 4. PubMed PMID: 24896178.

63. Meynert AM, Ansari M, FitzPatrick DR, Taylor MS. Variant detection sensitivity and biases in whole genome and exome sequencing. BMC Bioinformatics. 2014 Jul 19;15(1):247. PubMed PMID: 25038816.

64. Piton A, Redin C, Mandel JL. XLID-Causing Mutations and Associated Genes Challenged in Light of Data From Large-Scale Human Exome Sequencing. Am J Hum Genet. 2013 Jul 18. PubMed PMID: 23871722. Pubmed Central PMCID: 3738825.

65. Adzhubei IA, Schmidt S, Peshkin L, Ramensky VE, Gerasimova A, Bork P, et al. A method and server for predicting damaging missense mutations. Nat Methods. 2010 Apr;7(4):248-9. PubMed PMID: 20354512. Pubmed Central PMCID: 2855889.

66. Kumar P, Henikoff S, Ng PC. Predicting the effects of coding non-synonymous variants on protein function using the SIFT algorithm. Nat Protoc. 2009;4(7):1073-81. PubMed PMID: 19561590.

67. Cooper GM, Stone EA, Asimenos G, Program NCS, Green ED, Batzoglou S, et al. Distribution and intensity of constraint in mammalian genomic sequence. Genome Res. 2005 Jul;15(7):901-13. PubMed PMID: 15965027. Pubmed Central PMCID: 1172034.

68. Huang N, Lee I, Marcotte EM, Hurles ME. Characterising and predicting haploinsufficiency in the human genome. PLoS genetics. 2010 Oct;6(10):e1001154. PubMed PMID: 20976243. Pubmed Central PMCID: 2954820.

69. Aerts S, Lambrechts D, Maity S, Van Loo P, Coessens B, De Smet F, et al. Gene prioritization through genomic data fusion. Nat Biotechnol. 2006 May;24(5):537-44. PubMed PMID: 16680138. Epub 2006/05/09. eng.

70. Dan S, Chen F, Choy KW, Jiang F, Lin J, Xuan Z, et al. Prenatal detection of aneuploidy and imbalanced chromosomal arrangements by massively parallel sequencing. PLoS One. 2012;7(2):e27835. PubMed PMID: 22389664. Pubmed Central PMCID: 3289612.

71. Talkowski ME, Ordulu Z, Pillalamarri V, Benson CB, Blumenthal I, Connolly S, et al. Clinical diagnosis by whole-genome sequencing of a prenatal sample. N Engl J Med. 2012 Dec 6;367(23):2226-32. PubMed PMID: 23215558. Epub 2012/12/12. eng.

72. Filges I, Nosova E, Bruder E, Tercanli S, Townsend K, Gibson W, et al. Exome sequencing identifies mutations in KIF14 as a novel cause of an autosomal recessive lethal fetal ciliopathy phenotype. Clin Genet. 2013 Oct 15. PubMed PMID: 24128419.

73. Carss KJ, Hillman SC, Parthiban V, McMullan DJ, Maher ER, Kilby MD, et al. Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. Hum Mol Genet. 2014 Feb 11. PubMed PMID: 24476948.

74. Mackie FL, Carss KJ, Hillman SC, Hurles ME, Kilby MD. Exome sequencing in fetuses with structural malformations [review]. Journal of Clinical Medicine. 2014;3(3):747-62.

75. Robinson PN, Mundlos S. The human phenotype ontology. Clin Genet. 2010 Jun;77(6):525-34. PubMed PMID: 20412080.

76. Li H, Handsaker B, Wysoker A, Fennell T, Ruan J, Homer N, et al. The Sequence Alignment/Map format and SAMtools. Bioinformatics. 2009 Aug 15;25(16):2078-9. PubMed PMID: 19505943. Pubmed Central PMCID: 2723002.

77. Albers CA, Lunter G, MacArthur DG, McVean G, Ouwehand WH, Durbin R. Dindel: accurate indel calls from short-read data. Genome Res. 2011 Jun;21(6):961-73. PubMed PMID: 20980555. Pubmed Central PMCID: 3106329.

78. Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, et al. The variant call format and VCFtools. Bioinformatics. 2011 Aug 1;27(15):2156-8. PubMed PMID: 21653522. Pubmed Central PMCID: 3137218.

79. Flicek P, Ahmed I, Amode MR, Barrell D, Beal K, Brent S, et al. Ensembl 2013. Nucleic Acids Res. 2013 Jan;41(Database issue):D48-55. PubMed PMID: 23203987. Pubmed Central PMCID: 3531136.

80. Genomes Project Consortium, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, et al. An integrated map of genetic variation from 1,092 human genomes. Nature. 2012 Nov 1;491(7422):56-65. PubMed PMID: 23128226. Pubmed Central PMCID: 3498066.

81. Ramu A, Noordam MJ, Schwartz RS, Wuster A, Hurles ME, Cartwright RA, et al. DeNovoGear: de novo indel and point mutation discovery and phasing. Nat Methods. 2013 Oct;10(10):985-7. PubMed PMID: 23975140.

82. Thorvaldsdottir H, Robinson JT, Mesirov JP. Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. Brief Bioinform. 2013 Mar;14(2):178-92. PubMed PMID: 22517427. Pubmed Central PMCID: 3603213.

83. Kryukov GV, Pennacchio LA, Sunyaev SR. Most rare missense alleles are deleterious in humans: implications for complex disease and association studies. Am J Hum Genet. 2007 Apr;80(4):727-39. PubMed PMID: 17357078. Pubmed Central PMCID: 1852724. Epub 2007/03/16. eng.

84. Rauch A, Wieczorek D, Graf E, Wieland T, Endele S, Schwarzmayr T, et al. Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet. 2012 Sep 26. PubMed PMID: 23020937. Epub 2012/10/02. Eng.

85. Smigielski EM, Sirotkin K, Ward M, Sherry ST. dbSNP: a database of single nucleotide polymorphisms. Nucleic Acids Res. 2000 Jan 1;28(1):352-5. PubMed PMID: 10592272. Pubmed Central PMCID: 102496.

86. Dorschner MO, Amendola LM, Turner EH, Robertson PD, Shirts BH, Gallego CJ, et al. Actionable, pathogenic incidental findings in 1,000 participants' exomes. Am J Hum Genet. 2013 Oct 3;93(4):631-40. PubMed PMID: 24055113. Pubmed Central PMCID: 3791261.

87. Ku CS, Cooper DN, Polychronakos C, Naidoo N, Wu M, Soong R. Exome sequencing: dual role as a discovery and diagnostic tool. Ann Neurol. 2012 Jan;71(1):5-14. PubMed PMID: 22275248.

88. Bainbridge MN, Wang M, Wu Y, Newsham I, Muzny DM, Jefferies JL, et al. Targeted enrichment beyond the consensus coding DNA sequence exome reveals exons with higher variant densities. Genome Biol. 2011;12(7):R68. PubMed PMID: 21787409. Pubmed Central PMCID: 3218830.

89. Mills RE, Luttig CT, Larkins CE, Beauchamp A, Tsui C, Pittard WS, et al. An initial map of insertion and deletion (INDEL) variation in the human genome. Genome Res. 2006 Sep;16(9):1182-90. PubMed PMID: 16902084. Pubmed Central PMCID: 1557762.

90. Awadalla P, Gauthier J, Myers RA, Casals F, Hamdan FF, Griffing AR, et al. Direct measure of the de novo mutation rate in autism and schizophrenia cohorts. Am J Hum Genet. 2010 Sep 10;87(3):316-24. PubMed PMID: 20797689. Pubmed Central PMCID: 2933353. Epub 2010/08/28. eng.

91. Foldynova-Trantirkova S, Wilcox WR, Krejci P. Sixteen years and counting: the current understanding of fibroblast growth factor receptor 3 (FGFR3) signaling in skeletal dysplasias. Hum Mutat. 2012 Jan;33(1):29-41. PubMed PMID: 22045636. Pubmed Central PMCID: 3240715. Epub 2011/11/03. eng.

92. Potocki L, Abuelo DN, Oyer CE. Cardiac malformation in two infants with hypochondrogenesis. Am J Med Genet. 1995 Nov 20;59(3):295-9. PubMed PMID: 8599352. Epub 1995/11/20. eng.

93. Rittler M, Orioli IM. Achondrogenesis type II with polydactyly. Am J Med Genet. 1995 Nov 6;59(2):157-60. PubMed PMID: 8588578. Epub 1995/11/06. eng.

94. Zankl A, Zabel B, Hilbert K, Wildhardt G, Cuenot S, Xavier B, et al. Spondyloperipheral dysplasia is caused by truncating mutations in the C-propeptide of COL2A1. Am J Med Genet A. 2004 Aug 30;129A(2):144-8. PubMed PMID: 15316962. Epub 2004/08/19. eng.

95. Nishimura G, Haga N, Kitoh H, Tanaka Y, Sonoda T, Kitamura M, et al. The phenotypic spectrum of COL2A1 mutations. Hum Mutat. 2005 Jul;26(1):36-43. PubMed PMID: 15895462. Epub 2005/05/17. eng.

96. De Luca A, Bottillo I, Sarkozy A, Carta C, Neri C, Bellacchio E, et al. NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. Am J Hum Genet. 2005 Dec;77(6):1092-101. PubMed PMID: 16380919. Pubmed Central PMCID: 1285166. Epub 2005/12/29. eng.

97. Fahsold R, Hoffmeyer S, Mischung C, Gille C, Ehlers C, Kucukceylan N, et al. Minor lesion mutational spectrum of the entire NF1 gene does not explain its high mutability but points to a functional domain upstream of the GAP-related domain. Am J Hum Genet. 2000 Mar;66(3):790-818. PubMed PMID: 10712197. Pubmed Central PMCID: 1288164. Epub 2000/03/11. eng.

98. Padmanabhan A, Lee JS, Ismat FA, Lu MM, Lawson ND, Kanki JP, et al. Cardiac and vascular functions of the zebrafish orthologues of the type I neurofibromatosis gene NFI. Proc Natl Acad Sci U S A. 2009 Dec 29;106(52):22305-10. PubMed PMID: 19966217. Pubmed Central PMCID: 2799742. Epub 2009/12/08. eng.

99. Yan Z, Wang Z, Sharova L, Sharov AA, Ling C, Piao Y, et al. BAF250B-associated SWI/SNF chromatin-remodeling complex is required to maintain undifferentiated mouse embryonic stem cells. Stem Cells. 2008 May;26(5):1155-65. PubMed PMID: 18323406. Pubmed Central PMCID: 2409195. Epub 2008/03/08. eng.

100. Boerkoel CF, Takashima H, John J, Yan J, Stankiewicz P, Rosenbarker L, et al. Mutant chromatin remodeling protein SMARCAL1 causes Schimke immuno-osseous dysplasia. Nat Genet. 2002 Feb;30(2):215-20. PubMed PMID: 11799392. Epub 2002/01/19. eng.

101. Endele S, Rosenberger G, Geider K, Popp B, Tamer C, Stefanova I, et al. Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nat Genet. 2010 Nov;42(11):1021-6. PubMed PMID: 20890276. Epub 2010/10/05. eng.

102. Gao L, Macara IG, Joberty G. Multiple splice variants of Par3 and of a novel related gene, Par3L, produce proteins with different binding properties. Gene. 2002 Jul 10;294(1-2):99-107. PubMed PMID: 12234671.

103. Hong E, Jayachandran P, Brewster R. The polarity protein Pard3 is required for centrosome positioning during neurulation. Dev Biol. 2010 May 15;341(2):335-45. PubMed PMID: 20138861. Pubmed Central PMCID: 2862117.

104. Hirose T, Karasawa M, Sugitani Y, Fujisawa M, Akimoto K, Ohno S, et al. PAR3 is essential for cyst-mediated epicardial development by establishing apical cortical domains. Development. 2006 Apr;133(7):1389-98. PubMed PMID: 16510507.

105. Bisschoff IJ, Zeschnigk C, Horn D, Wellek B, Riess A, Wessels M, et al. Novel mutations including deletions of the entire OFD1 gene in 30 families with type 1 orofaciodigital syndrome: a study of the extensive clinical variability. Hum Mutat. 2013 Jan;34(1):237-47. PubMed PMID: 23033313. Epub 2012/10/04. eng.

106. Durkin ME, Avner MR, Huh CG, Yuan BZ, Thorgeirsson SS, Popescu NC. DLC-1, a Rho GTPase-activating protein with tumor suppressor function, is essential for embryonic development. FEBS Lett. 2005 Feb 14;579(5):1191-6. PubMed PMID: 15710412. Epub 2005/02/16. eng.

107. Vilhais-Neto GC, Maruhashi M, Smith KT, Vasseur-Cognet M, Peterson AS, Workman JL, et al. Rere controls retinoic acid signalling and somite bilateral symmetry. Nature. 2010 Feb 18;463(7283):953-7. PubMed PMID: 20164929. Epub 2010/02/19. eng.

108. Plaster N, Sonntag C, Schilling TF, Hammerschmidt M. REREa/Atrophin-2 interacts with histone deacetylase and Fgf8 signaling to regulate multiple processes of zebrafish development. Dev Dyn. 2007 Jul;236(7):1891-904. PubMed PMID: 17576618. Epub 2007/06/20. eng.

109. Kim BJ, Zaveri HP, Shchelochkov OA, Yu Z, Hernandez-Garcia A, Seymour ML, et al. An allelic series of mice reveals a role for RERE in the development of multiple organs affected in chromosome 1p36 deletions. PLoS One. 2013;8(2):e57460. PubMed PMID: 23451234. Pubmed Central PMCID: 3581587. Epub 2013/03/02. eng.

110. Liu W, Morito D, Takashima S, Mineharu Y, Kobayashi H, Hitomi T, et al. Identification of RNF213 as a susceptibility gene for moyamoya disease and its possible role in vascular development. PLoS One. 2011;6(7):e22542. PubMed PMID: 21799892. Pubmed Central PMCID: 3140517. Epub 2011/07/30. eng.

111. Kida Y, Maeda Y, Shiraishi T, Suzuki T, Ogura T. Chick Dach1 interacts with the Smad complex and Sin3a to control AER formation and limb development along the proximodistal axis. Development. 2004 Sep;131(17):4179-87. PubMed PMID: 15280207. Epub 2004/07/29. eng.

112. Martini SR, Davis RL. The dachshund gene is required for the proper guidance and branching of mushroom body axons in Drosophila melanogaster. J Neurobiol. 2005 Aug;64(2):133-44. PubMed PMID: 15818552. Epub 2005/04/09. eng.

113. Backman M, Machon O, Van Den Bout CJ, Krauss S. Targeted disruption of mouse Dach1 results in postnatal lethality. Dev Dyn. 2003 Jan;226(1):139-44. PubMed PMID: 12508235. Epub 2003/01/01. eng.

114. Boyd PA, Keeling JW, Lindenbaum RH. Fraser syndrome (cryptophthalmos-syndactyly syndrome): a review of eleven cases with postmortem findings. Am J Med Genet. 1988 Sep;31(1):159-68. PubMed PMID: 2851937. Epub 1988/09/01. eng.

115. Pitera JE, Scambler PJ, Woolf AS. Fras1, a basement membrane-associated protein mutated in Fraser syndrome, mediates both the initiation of the mammalian kidney and the integrity of renal glomeruli. Hum Mol Genet. 2008 Dec 15;17(24):3953-64. PubMed PMID: 18787044. Pubmed Central PMCID: 2638576. Epub 2008/09/13. eng.

116. Kerecuk L, Long DA, Ali Z, Anders C, Kolatsi-Joannou M, Scambler PJ, et al. Expression of Fraser syndrome genes in normal and polycystic murine kidneys. Pediatr Nephrol. 2012 Feb 1. PubMed PMID: 22294133. Epub 2012/02/02. Eng.

117. Vrontou S, Petrou P, Meyer BI, Galanopoulos VK, Imai K, Yanagi M, et al. Fras1 deficiency results in cryptophthalmos, renal agenesis and blebbed phenotype in mice. Nat Genet. 2003 Jun;34(2):209-14. PubMed PMID: 12766770.

118. Talbot JC, Walker MB, Carney TJ, Huycke TR, Yan YL, BreMiller RA, et al. fras1 shapes endodermal pouch 1 and stabilizes zebrafish pharyngeal skeletal development. Development. 2012 Aug;139(15):2804-13. PubMed PMID: 22782724. Pubmed Central PMCID: 3392706. Epub 2012/07/12. eng.

119. Gautier P, Naranjo-Golborne C, Taylor MS, Jackson IJ, Smyth I. Expression of the fras1/frem gene family during zebrafish development and fin morphogenesis. Dev Dyn. 2008 Nov;237(11):3295-304. PubMed PMID: 18816440. Epub 2008/09/26. eng.

120. Sifrim A, Popovic D, Tranchevent LC, Ardeshirdavani A, Sakai R, Konings P, et al. eXtasy: variant prioritization by genomic data fusion. Nat Methods. 2013 Nov;10(11):1083-4. PubMed PMID: 24076761.

121. Smedley D, Oellrich A, Kohler S, Ruef B, Sanger Mouse Genetics P, Westerfield M, et al. PhenoDigm: analyzing curated annotations to associate animal models with human diseases. Database (Oxford). 2013;2013:bat025. PubMed PMID: 23660285. Pubmed Central PMCID: 3649640.

122. Woodbine L, Neal JA, Sasi NK, Shimada M, Deem K, Coleman H, et al. PRKDC mutations in a SCID patient with profound neurological abnormalities. J Clin Invest. 2013 Jul 1;123(7):2969-80. PubMed PMID: 23722905.

123. Goryunov D, He CZ, Lin CS, Leung CL, Liem RK. Nervous-tissue-specific elimination of microtubule-actin crosslinking factor 1a results in multiple developmental defects in the mouse brain. Mol Cell Neurosci. 2010 May;44(1):1-14. PubMed PMID: 20170731. Pubmed Central PMCID: 2847646.

124. Mefford HC, Clauin S, Sharp AJ, Moller RS, Ullmann R, Kapur R, et al. Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. Am J Hum Genet. 2007 Nov;81(5):1057-69. PubMed PMID: 17924346. Pubmed Central PMCID: 2265663. Epub 2007/10/10. eng.

125. Liao BY, Zhang J. Null mutations in human and mouse orthologs frequently result in different phenotypes. Proc Natl Acad Sci U S A. 2008 May 13;105(19):6987-92. PubMed PMID: 18458337. Pubmed Central PMCID: 2383943.

126. Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, Martin CL, et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genet Med. 2013 Jul;15(7):565-74. PubMed PMID: 23788249. Pubmed Central PMCID: 3727274.

127. Bernhardt BA, Soucier D, Hanson K, Savage MS, Jackson L, Wapner RJ. Women's experiences receiving abnormal prenatal chromosomal microarray testing results. Genet Med. 2013 Feb;15(2):139-45. PubMed PMID: 22955112. Pubmed Central PMCID: 3877835.

128. McGillivray G, Rosenfeld JA, McKinlay Gardner RJ, Gillam LH. Genetic counselling and ethical issues with chromosome microarray analysis in prenatal testing. Prenat Diagn. 2012 Apr;32(4):389-95. PubMed PMID: 22467169.

129. Ford D, Easton DF, Stratton M, Narod S, Goldgar D, Devilee P, et al. Genetic heterogeneity and penetrance analysis of the BRCA1 and BRCA2 genes in breast cancer families. The Breast Cancer Linkage Consortium. Am J Hum Genet. 1998 Mar;62(3):676-89. PubMed PMID: 9497246. Pubmed Central PMCID: 1376944.

130. Yurkiewicz IR, Korf BR, Lehmann LS. Prenatal whole-genome sequencing--is the quest to know a fetus's future ethical? N Engl J Med. 2014 Jan 16;370(3):195-7. PubMed PMID: 24428465.

131. Weedon MN, Cebola I, Patch AM, Flanagan SE, De Franco E, Caswell R, et al. Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nat Genet. 2013 Nov 10. PubMed PMID: 24212882.

132. Dathe K, Kjaer KW, Brehm A, Meinecke P, Nurnberg P, Neto JC, et al. Duplications involving a conserved regulatory element downstream of BMP2 are associated with brachydactyly type A2. Am J Hum Genet. 2009 Apr;84(4):483-92. PubMed PMID: 19327734. Pubmed Central PMCID: 2667973.

133. Biesecker LG, Shianna KV, Mullikin JC. Exome sequencing: the expert view. Genome Biol. 2011;12(9):128. PubMed PMID: 21920051. Pubmed Central PMCID: 3308041.

134. Press release: DNA tests to revolutionise fight against cancer and help 100,000 NHS patients 2012. Available from: https://www.gov.uk/government/news/dna-tests-to-revolutionise-fight-against-cancer-and-help-100000-nhs-patients.

135. van Bokhoven H. Genetic and epigenetic networks in intellectual disabilities. Annu Rev Genet. 2011;45:81-104. PubMed PMID: 21910631.

136. Bryson SE, Bradley EA, Thompson A, Wainwright A. Prevalence of autism among adolescents with intellectual disabilities. Can J Psychiatry. 2008 Jul;53(7):449-59. PubMed PMID: 18674403.

137. Kaufman L, Ayub M, Vincent JB. The genetic basis of non-syndromic intellectual disability: a review. J Neurodev Disord. 2010 Dec;2(4):182-209. PubMed PMID: 21124998. Pubmed Central PMCID: 2974911.

138. Leonard H, Wen X. The epidemiology of mental retardation: challenges and opportunities in the new millennium. Mental retardation and developmental disabilities research reviews. 2002;8(3):117-34. PubMed PMID: 12216056.

139. Maulik PK, Mascarenhas MN, Mathers CD, Dua T, Saxena S. Prevalence of intellectual disability: a meta-analysis of population-based studies. Res Dev Disabil. 2011 Mar-Apr;32(2):419-36. PubMed PMID: 21236634.

140. Baird PA, Sadovnick AD. Mental retardation in over half-a-million consecutive livebirths: an epidemiological study. Am J Ment Defic. 1985 Jan;89(4):323-30. PubMed PMID: 3976730.

141. Centers for Disease C, Prevention. Economic costs associated with mental retardation, cerebral palsy, hearing loss, and vision impairment--United States, 2003. MMWR Morb Mortal Wkly Rep. 2004 Jan 30;53(3):57-9. PubMed PMID: 14749614.

142. Waber DP, Bryce CP, Girard JM, Zichlin M, Fitzmaurice GM, Galler JR. Impaired IQ and academic skills in adults who experienced moderate to severe infantile malnutrition: a 40-year study. Nutr Neurosci. 2014 Feb;17(2):58-64. PubMed PMID: 23484464. Pubmed Central PMCID: 3796166.

143. Niccols A. Fetal alcohol syndrome and the developing socio-emotional brain. Brain Cogn. 2007 Oct;65(1):135-42. PubMed PMID: 17669569.

144. Freij BJ, South MA, Sever JL. Maternal rubella and the congenital rubella syndrome. Clin Perinatol. 1988 Jun;15(2):247-57. PubMed PMID: 3288422.

145. Solon O, Riddell TJ, Quimbo SA, Butrick E, Aylward GP, Lou Bacate M, et al. Associations between cognitive function, blood lead concentration, and nutrition among children in the central Philippines. J Pediatr. 2008 Feb;152(2):237-43. PubMed PMID: 18206696.

146. Seidman LJ, Buka SL, Goldstein JM, Horton NJ, Rieder RO, Tsuang MT. The relationship of prenatal and perinatal complications to cognitive functioning at age 7 in the New England Cohorts of the National Collaborative Perinatal Project. Schizophr Bull. 2000;26(2):309-21. PubMed PMID: 10885633.

147. L. P. A clinical and genetic study of 1280 cases of mental defect. London: HMSO. 1938;229.

148. E. S. Mental Deficiency. The Eugenics Review. 1938;30(3):208-9.

149. Haldane JBS. A Clinical and Genetic Study of 1280 Cases of Mental Defect. Nature. 1938;141:575-6.

150. Deary IJ, Johnson W, Houlihan LM. Genetic foundations of human intelligence. Hum Genet. 2009 Jul;126(1):215-32. PubMed PMID: 19294424.

151. Rietveld CA, Medland SE, Derringer J, Yang J, Esko T, Martin NW, et al. GWAS of 126,559 individuals identifies genetic variants associated with educational attainment. Science. 2013 Jun 21;340(6139):1467-71. PubMed PMID: 23722424. Pubmed Central PMCID: 3751588.

152. Rauch A, Hoyer J, Guth S, Zweier C, Kraus C, Becker C, et al. Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. Am J Med Genet A. 2006 Oct 1;140(19):2063-74. PubMed PMID: 16917849.

153. Lejeune J, Turpin R, Gautier M. [Mongolism; a chromosomal disease (trisomy)]. Bull Acad Natl Med. 1959 Apr 7-14;143(11-12):256-65. PubMed PMID: 13662687. Le mongolisme, maladie chromosomique. (trisomie).

154. Shaw-Smith C, Pittman AM, Willatt L, Martin H, Rickman L, Gribble S, et al. Microdeletion encompassing MAPT at chromosome 17q21.3 is associated with developmental delay and learning disability. Nat Genet. 2006 Sep;38(9):1032-7. PubMed PMID: 16906163.

155. Cooper GM, Coe BP, Girirajan S, Rosenfeld JA, Vu TH, Baker C, et al. A copy number variation morbidity map of developmental delay. Nat Genet. 2011 Aug 14. PubMed PMID: 21841781. Eng.

156. Talkowski ME, Mullegama SV, Rosenfeld JA, van Bon BW, Shen Y, Repnikova EA, et al. Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. Am J Hum Genet. 2011 Oct 7;89(4):551-63. PubMed PMID: 21981781. Pubmed Central PMCID: 3188839.

157. Zollino M, Orteschi D, Murdolo M, Lattante S, Battaglia D, Stefanini C, et al. Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome phenotype. Nat Genet. 2012 Jun;44(6):636-8. PubMed PMID: 22544367.

158. Girirajan S, Rosenfeld JA, Cooper GM, Antonacci F, Siswara P, Itsara A, et al. A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nat Genet. 2010 Mar;42(3):203-9. PubMed PMID: 20154674. Pubmed Central PMCID: 2847896.

159. Verkerk AJ, Pieretti M, Sutcliffe JS, Fu YH, Kuhl DP, Pizzuti A, et al. Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint

cluster region exhibiting length variation in fragile X syndrome. Cell. 1991 May 31;65(5):905-14. PubMed PMID: 1710175.

160. Coffee B, Keith K, Albizua I, Malone T, Mowrey J, Sherman SL, et al. Incidence of fragile X syndrome by newborn screening for methylated FMR1 DNA. Am J Hum Genet. 2009 Oct;85(4):503-14. PubMed PMID: 19804849. Pubmed Central PMCID: 2756550.

161. Ausio J, Paz AM, Esteller M. MeCP2: the long trip from a chromatin protein to neurological disorders. Trends Mol Med. 2014 Apr 21. PubMed PMID: 24766768.

162. Ropers HH. Genetics of early onset cognitive impairment. Annu Rev Genomics Hum Genet. 2010 Sep 22;11:161-87. PubMed PMID: 20822471.

163. Najmabadi H, Hu H, Garshasbi M, Zemojtel T, Abedini SS, Chen W, et al. Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature. 2011 Oct 6;478(7367):57-63. PubMed PMID: 21937992. Epub 2011/09/23. eng.

164. Vissers LE, de Ligt J, Gilissen C, Janssen I, Steehouwer M, de Vries P, et al. A de novo paradigm for mental retardation. Nat Genet. 2010 Dec;42(12):1109-12. PubMed PMID: 21076407.

165. Salvador-Carulla L, Bertelli M. 'Mental retardation' or 'intellectual disability': time for a conceptual change. Psychopathology. 2008;41(1):10-6. PubMed PMID: 17952016.

166. Saitsu H, Kato M, Mizuguchi T, Hamada K, Osaka H, Tohyama J, et al. De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nat Genet. 2008 Jun;40(6):782-8. PubMed PMID: 18469812.

167. Zhang Y, Luan Z, Liu A, Hu G. The scaffolding protein CASK mediates the interaction between rabphilin3a and beta-neurexins. FEBS Lett. 2001 May 25;497(2-3):99-102. PubMed PMID: 11377421.

168. Carrie A, Jun L, Bienvenu T, Vinet MC, McDonell N, Couvert P, et al. A new member of the IL-1 receptor family highly expressed in hippocampus and involved in X-linked mental retardation. Nat Genet. 1999 Sep;23(1):25-31. PubMed PMID: 10471494.

169. Wu Y, Arai AC, Rumbaugh G, Srivastava AK, Turner G, Hayashi T, et al. Mutations in ionotropic AMPA receptor 3 alter channel properties and are associated

with moderate cognitive impairment in humans. Proc Natl Acad Sci U S A. 2007 Nov 13;104(46):18163-8. PubMed PMID: 17989220. Pubmed Central PMCID: 2084314.

170. Hamdan FF, Gauthier J, Spiegelman D, Noreau A, Yang Y, Pellerin S, et al. Mutations in SYNGAP1 in autosomal nonsyndromic mental retardation. N Engl J Med. 2009 Feb 5;360(6):599-605. PubMed PMID: 19196676. Pubmed Central PMCID: 2925262.

171. Berkel S, Marshall CR, Weiss B, Howe J, Roeth R, Moog U, et al. Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. Nat Genet. 2010 Jun;42(6):489-91. PubMed PMID: 20473310.

172. Kleefstra T, Smidt M, Banning MJ, Oudakker AR, Van Esch H, de Brouwer AP, et al. Disruption of the gene Euchromatin Histone Methyl Transferase1 (Eu-HMTase1) is associated with the 9q34 subtelomeric deletion syndrome. J Med Genet. 2005 Apr;42(4):299-306. PubMed PMID: 15805155. Pubmed Central PMCID: 1736026.

173. Hoyer J, Ekici AB, Endele S, Popp B, Zweier C, Wiesener A, et al. Haploinsufficiency of ARID1B, a member of the SWI/SNF-a chromatin-remodeling complex, is a frequent cause of intellectual disability. Am J Hum Genet. 2012 Mar 9;90(3):565-72. PubMed PMID: 22405089. Pubmed Central PMCID: 3309205. Epub 2012/03/13. eng.

174. Zalfa F, Eleuteri B, Dickson KS, Mercaldo V, De Rubeis S, di Penta A, et al. A new function for the fragile X mental retardation protein in regulation of PSD-95 mRNA stability. Nat Neurosci. 2007 May;10(5):578-87. PubMed PMID: 17417632. Pubmed Central PMCID: 2804293.

175. Paine RS. The variability in manifestations of untreated patients with phenylketonuria (phenylpyruvic aciduria). Pediatrics. 1957 Aug;20(2):290-302. PubMed PMID: 13452670.

176. de Lonlay P, Seta N, Barrot S, Chabrol B, Drouin V, Gabriel BM, et al. A broad spectrum of clinical presentations in congenital disorders of glycosylation I: a series of 26 cases. J Med Genet. 2001 Jan;38(1):14-9. PubMed PMID: 11134235. Pubmed Central PMCID: 1734729.

177. Fiala JC, Spacek J, Harris KM. Dendritic spine pathology: cause or consequence of neurological disorders? Brain Res Brain Res Rev. 2002 Jun;39(1):29-54. PubMed PMID: 12086707.

178. Zanni G, Saillour Y, Nagara M, Billuart P, Castelnau L, Moraine C, et al. Oligophrenin 1 mutations frequently cause X-linked mental retardation with cerebellar hypoplasia. Neurology. 2005 Nov 8;65(9):1364-9. PubMed PMID: 16221952.

179. Lebel RR, May M, Pouls S, Lubs HA, Stevenson RE, Schwartz CE. Non-syndromic X-linked mental retardation associated with a missense mutation (P312L) in the FGD1 gene. Clin Genet. 2002 Feb;61(2):139-45. PubMed PMID: 11940089.

180. Kerr B, Delrue MA, Sigaudy S, Perveen R, Marche M, Burgelin I, et al. Genotype-phenotype correlation in Costello syndrome: HRAS mutation analysis in 43 cases. J Med Genet. 2006 May;43(5):401-5. PubMed PMID: 16443854. Pubmed Central PMCID: 2564514.

181. San Martin A, Pagani MR. Understanding intellectual disability through RASopathies. J Physiol Paris. 2014 May 21. PubMed PMID: 24859216.

182. Barrientos RM, O'Reilly RC, Rudy JW. Memory for context is impaired by injecting anisomycin into dorsal hippocampus following context exploration. Behav Brain Res. 2002 Aug 21;134(1-2):299-306. PubMed PMID: 12191817.

183. Ohno H, Shinoda K, Ohyama K, Sharp LZ, Kajimura S. EHMT1 controls brown adipose cell fate and thermogenesis through the PRDM16 complex. Nature. 2013 Dec 5;504(7478):163-7. PubMed PMID: 24196706. Pubmed Central PMCID: 3855638.

184. Lee S, Abecasis GR, Boehnke M, Lin X. Rare-Variant Association Analysis: Study Designs and Statistical Tests. Am J Hum Genet. 2014 Jul 3;95(1):5-23. PubMed PMID: 24995866. Pubmed Central PMCID: 4085641.

185. Morgenthaler S, Thilly WG. A strategy to discover genes that carry multi-allelic or mono-allelic risk for common diseases: a cohort allelic sums test (CAST). Mutat Res. 2007 Feb 3;615(1-2):28-56. PubMed PMID: 17101154.

186. Cohen JC, Kiss RS, Pertsemlidis A, Marcel YL, McPherson R, Hobbs HH. Multiple rare alleles contribute to low plasma levels of HDL cholesterol. Science. 2004 Aug 6;305(5685):869-72. PubMed PMID: 15297675.

187. Madsen BE, Browning SR. A groupwise association test for rare mutations using a weighted sum statistic. PLoS genetics. 2009 Feb;5(2):e1000384. PubMed PMID: 19214210. Pubmed Central PMCID: 2633048.

188. Wu MC, Lee S, Cai T, Li Y, Boehnke M, Lin X. Rare-variant association testing for sequencing data with the sequence kernel association test. Am J Hum Genet. 2011 Jul 15;89(1):82-93. PubMed PMID: 21737059. Pubmed Central PMCID: 3135811.

189. Cardon LR, Palmer LJ. Population stratification and spurious allelic association. Lancet. 2003 Feb 15;361(9357):598-604. PubMed PMID: 12598158.

190. Liu L, Sabo A, Neale BM, Nagaswamy U, Stevens C, Lim E, et al. Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. PLoS genetics. 2013 Apr;9(4):e1003443. PubMed PMID: 23593035. Pubmed Central PMCID: 3623759.

191. Grozeva D, Carss K, Spasic-Boskovic O, Parker MJ, Archer H, Firth HV, et al. De novo loss-of-function mutations in SETD5, encoding a methyltransferase in a 3p25 microdeletion syndrome critical region, cause intellectual disability. Am J Hum Genet. 2014 Apr 3;94(4):618-24. PubMed PMID: 24680889. Pubmed Central PMCID: 3980521.

192. McKenna A, Hanna M, Banks E, Sivachenko A, Cibulskis K, Kernytsky A, et al. The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res. 2010 Sep;20(9):1297-303. PubMed PMID: 20644199. Pubmed Central PMCID: 2928508. Epub 2010/07/21. eng.

193. McLaren W, Pritchard B, Rios D, Chen Y, Flicek P, Cunningham F. Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. Bioinformatics. 2010 Aug 15;26(16):2069-70. PubMed PMID: 20562413. Pubmed Central PMCID: 2916720. Epub 2010/06/22. eng.

194. Zheng X, Levine D, Shen J, Gogarten SM, Laurie C, Weir BS. A high-performance computing toolset for relatedness and principal component analysis of SNP data. Bioinformatics. 2012 Dec 15;28(24):3326-8. PubMed PMID: 23060615. Pubmed Central PMCID: 3519454.

195. International HapMap C, Altshuler DM, Gibbs RA, Peltonen L, Altshuler DM, Gibbs RA, et al. Integrating common and rare genetic variation in diverse human populations. Nature. 2010 Sep 2;467(7311):52-8. PubMed PMID: 20811451. Pubmed Central PMCID: 3173859.

196. Gonzalez-Perez A, Lopez-Bigas N. Improving the assessment of the outcome of nonsynonymous SNVs with a consensus deleteriousness score, Condel. Am J Hum

Genet. 2011 Apr 8;88(4):440-9. PubMed PMID: 21457909. Pubmed Central PMCID: 3071923.

197. Iossifov I, Ronemus M, Levy D, Wang Z, Hakker I, Rosenbaum J, et al. De novo gene disruptions in children on the autistic spectrum. Neuron. 2012 Apr 26;74(2):285-99. PubMed PMID: 22542183. Pubmed Central PMCID: 3619976.

198. Neale BM, Kou Y, Liu L, Ma'ayan A, Samocha KE, Sabo A, et al. Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature. 2012 May 10;485(7397):242-5. PubMed PMID: 22495311. Pubmed Central PMCID: 3613847.

199. Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, Klei L, et al. Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. Am J Hum Genet. 2014 May 1;94(5):677-94. PubMed PMID: 24768552. Pubmed Central PMCID: 4067558.

200. Kellogg G, Sum J, Wallerstein R. Deletion of 3p25.3 in a patient with intellectual disability and dysmorphic features with further definition of a critical region. Am J Med Genet A. 2013 Jun;161(6):1405-8. PubMed PMID: 23613140.

201. Verjaal M, De Nef MB. A patient with a partial deletion of the short arm of chromosome 3. Am J Dis Child. 1978 Jan;132(1):43-5. PubMed PMID: 623063.

202. Gunnarsson C, Foyn Bruun C. Molecular characterization and clinical features of a patient with an interstitial deletion of 3p25.3-p26.1. Am J Med Genet A. 2010 Dec;152A(12):3110-4. PubMed PMID: 21082655.

203. Riess A, Grasshoff U, Schaferhoff K, Bonin M, Riess O, Horber V, et al. Interstitial 3p25.3-p26.1 deletion in a patient with intellectual disability. Am J Med Genet A. 2012 Oct;158A(10):2587-90. PubMed PMID: 22965684.

204. Peltekova IT, Macdonald A, Armour CM. Microdeletion on 3p25 in a patient with features of 3p deletion syndrome. Am J Med Genet A. 2012 Oct;158A(10):2583-6. PubMed PMID: 22903836.

205. UniProt C. Update on activities at the Universal Protein Resource (UniProt) in 2013. Nucleic Acids Res. 2013 Jan;41(Database issue):D43-7. PubMed PMID: 23161681. Pubmed Central PMCID: 3531094.

206. Cao R, Wang L, Wang H, Xia L, Erdjument-Bromage H, Tempst P, et al. Role of histone H3 lysine 27 methylation in Polycomb-group silencing. Science. 2002 Nov 1;298(5595):1039-43. PubMed PMID: 12351676.

207. Plath K, Fang J, Mlynarczyk-Evans SK, Cao R, Worringer KA, Wang H, et al. Role of histone H3 lysine 27 methylation in X inactivation. Science. 2003 Apr 4;300(5616):131-5. PubMed PMID: 12649488.

208. Kamminga LM, Bystrykh LV, de Boer A, Houwer S, Douma J, Weersing E, et al. The Polycomb group gene Ezh2 prevents hematopoietic stem cell exhaustion. Blood. 2006 Mar 1;107(5):2170-9. PubMed PMID: 16293602. Pubmed Central PMCID: 1895717.

209. Weaver DD, Graham CB, Thomas IT, Smith DW. A new overgrowth syndrome with accelerated skeletal maturation, unusual facies, and camptodactyly. J Pediatr. 1974 Apr;84(4):547-52. PubMed PMID: 4366187.

210. Ng SB, Bigham AW, Buckingham KJ, Hannibal MC, McMillin MJ, Gildersleeve HI, et al. Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nat Genet. 2010 Sep;42(9):790-3. PubMed PMID: 20711175. Pubmed Central PMCID: 2930028.

211. Zechner U, Wilda M, Kehrer-Sawatzki H, Vogel W, Fundele R, Hameister H. A high density of X-linked genes for general cognitive ability: a run-away process shaping human evolution? Trends Genet. 2001 Dec;17(12):697-701. PubMed PMID: 11718922.

212. Nguyen DK, Disteche CM. High expression of the mammalian X chromosome in brain. Brain Res. 2006 Dec 18;1126(1):46-9. PubMed PMID: 16978591.

213. Deng X, Hiatt JB, Nguyen DK, Ercan S, Sturgill D, Hillier LW, et al. Evidence for compensatory upregulation of expressed X-linked genes in mammals, Caenorhabditis elegans and Drosophila melanogaster. Nat Genet. 2011 Dec;43(12):1179-85. PubMed PMID: 22019781. Pubmed Central PMCID: 3576853.

214. Hammer MF, Woerner AE, Mendez FL, Watkins JC, Cox MP, Wall JD. The ratio of human X chromosome to autosome diversity is positively correlated with genetic distance from genes. Nat Genet. 2010 Oct;42(10):830-1. PubMed PMID: 20802480.

215. Schaffner SF. The X chromosome in population genetics. Nature reviews Genetics. 2004 Jan;5(1):43-51. PubMed PMID: 14708015.

216. Beltran-Valero de Bernabe D, Currier S, Steinbrecher A, Celli J, van Beusekom E, van der Zwaag B, et al. Mutations in the O-mannosyltransferase gene POMT1 give rise to the severe neuronal migration disorder Walker-Warburg syndrome. Am J Hum

Genet. 2002 Nov;71(5):1033-43. PubMed PMID: 12369018. Pubmed Central PMCID: 419999. Epub 2002/10/09. eng.

217. Yoshida A, Kobayashi K, Manya H, Taniguchi K, Kano H, Mizuno M, et al. Muscular dystrophy and neuronal migration disorder caused by mutations in a glycosyltransferase, POMGnT1. Dev Cell. 2001 Nov;1(5):717-24. PubMed PMID: 11709191. Epub 2001/11/16. eng.

218. van Reeuwijk J, Maugenre S, van den Elzen C, Verrips A, Bertini E, Muntoni F, et al. The expanding phenotype of POMT1 mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. Hum Mutat. 2006 May;27(5):453-9. PubMed PMID: 16575835.

219. Godfrey C, Clement E, Mein R, Brockington M, Smith J, Talim B, et al. Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain : a journal of neurology. 2007 Oct;130(Pt 10):2725-35. PubMed PMID: 17878207. Epub 2007/09/20. eng.

220. Ibraghimov-Beskrovnaya O, Ervasti JM, Leveille CJ, Slaughter CA, Sernett SW, Campbell KP. Primary structure of dystrophin-associated glycoproteins linking dystrophin to the extracellular matrix. Nature. 1992 Feb 20;355(6362):696-702. PubMed PMID: 1741056. Epub 1992/02/20. eng.

221. van Reeuwijk J, Janssen M, van den Elzen C, Beltran-Valero de Bernabe D, Sabatelli P, Merlini L, et al. POMT2 mutations cause alpha-dystroglycan hypoglycosylation and Walker-Warburg syndrome. J Med Genet. 2005 Dec;42(12):907-12. PubMed PMID: 15894594. Pubmed Central PMCID: 1735967. Epub 2005/05/17. eng.

222. Wright KM, Lyon KA, Leung H, Leahy DJ, Ma L, Ginty DD. Dystroglycan organizes axon guidance cue localization and axonal pathfinding. Neuron. 2012 Dec 6;76(5):931-44. PubMed PMID: 23217742. Pubmed Central PMCID: 3526105. Epub 2012/12/12. eng.

223. Wells L. The o-mannosylation pathway: glycosyltransferases and proteins implicated in congenital muscular dystrophy. J Biol Chem. 2013 Mar 8;288(10):6930-5. PubMed PMID: 23329833. Pubmed Central PMCID: 3591603.

224. Stalnaker SH, Hashmi S, Lim JM, Aoki K, Porterfield M, Gutierrez-Sanchez G, et al. Site mapping and characterization of O-glycan structures on alpha-dystroglycan

isolated from rabbit skeletal muscle. J Biol Chem. 2010 Aug 6;285(32):24882-91. PubMed PMID: 20507986. Pubmed Central PMCID: 2915724. Epub 2010/05/29. eng.

225. Tran DT, Lim JM, Liu M, Stalnaker SH, Wells L, Ten Hagen KG, et al. Glycosylation of alpha-dystroglycan: O-mannosylation influences the subsequent addition of GalNAc by UDP-GalNAc polypeptide N-acetylgalactosaminyltransferases. J Biol Chem. 2012 Jun 15;287(25):20967-74. PubMed PMID: 22549772. Pubmed Central PMCID: 3375520.

226. Michele DE, Barresi R, Kanagawa M, Saito F, Cohn RD, Satz JS, et al. Post-translational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies. Nature. 2002 Jul 25;418(6896):417-22. PubMed PMID: 12140558. eng.

227. Yoshida-Moriguchi T, Yu L, Stalnaker SH, Davis S, Kunz S, Madson M, et al. O-mannosyl phosphorylation of alpha-dystroglycan is required for laminin binding. Science. 2010 Jan 1;327(5961):88-92. PubMed PMID: 20044576. Pubmed Central PMCID: 2978000. Epub 2010/01/02. eng.

228. Frost AR, Bohm SV, Sewduth RN, Josifova D, Ogilvie CM, Izatt L, et al. Heterozygous deletion of a 2-Mb region including the dystroglycan gene in a patient with mild myopathy, facial hypotonia, oral-motor dyspraxia and white matter abnormalities. Eur J Hum Genet. 2010 Jul;18(7):852-5. PubMed PMID: 20234391. Pubmed Central PMCID: 2987357.

229. Hara Y, Balci-Hayta B, Yoshida-Moriguchi T, Kanagawa M, Beltran-Valero de Bernabe D, Gundesli H, et al. A dystroglycan mutation associated with limb-girdle muscular dystrophy. N Engl J Med. 2011 Mar 10;364(10):939-46. PubMed PMID: 21388311. Pubmed Central PMCID: 3071687. Epub 2011/03/11. eng.

230. Geis T, Marquard K, Rodl T, Reihle C, Schirmer S, von Kalle T, et al. Homozygous dystroglycan mutation associated with a novel muscle-eye-brain disease-like phenotype with multicystic leucodystrophy. Neurogenetics. 2013 Nov;14(3-4):205-13. PubMed PMID: 24052401.

231. Maeda Y, Tanaka S, Hino J, Kangawa K, Kinoshita T. Human dolichol-phosphate-mannose synthase consists of three subunits, DPM1, DPM2 and DPM3. EMBO J. 2000 Jun 1;19(11):2475-82. PubMed PMID: 10835346. Pubmed Central PMCID: 212771.

232. Ashida H, Maeda Y, Kinoshita T. DPM1, the catalytic subunit of dolichol-phosphate mannose synthase, is tethered to and stabilized on the endoplasmic

reticulum membrane by DPM3. J Biol Chem. 2006 Jan 13;281(2):896-904. PubMed PMID: 16280320.

233. Kim S, Westphal V, Srikrishna G, Mehta DP, Peterson S, Filiano J, et al. Dolichol phosphate mannose synthase (DPM1) mutations define congenital disorder of glycosylation Ie (CDG-Ie). J Clin Invest. 2000 Jan;105(2):191-8. PubMed PMID: 10642597. Pubmed Central PMCID: 377427.

234. Lefeber DJ, Schonberger J, Morava E, Guillard M, Huyben KM, Verrijp K, et al. Deficiency of Dol-P-Man synthase subunit DPM3 bridges the congenital disorders of glycosylation with the dystroglycanopathies. Am J Hum Genet. 2009 Jul;85(1):76-86. PubMed PMID: 19576565. eng.

235. Barone R, Aiello C, Race V, Morava E, Foulquier F, Riemersma M, et al. DPM2-CDG: A muscular dystrophy-dystroglycanopathy syndrome with severe epilepsy. Ann Neurol. 2012 Oct;72(4):550-8. PubMed PMID: 23109149. Epub 2012/10/31. eng.

236. Yang AC, Ng BG, Moore SA, Rush J, Waechter CJ, Raymond KM, et al. Congenital disorder of glycosylation due to DPM1 mutations presenting with dystroglycanopathy-type congenital muscular dystrophy. Mol Genet Metab. 2013 Nov;110(3):345-51. PubMed PMID: 23856421. Pubmed Central PMCID: 3800268.

237. Lefeber DJ, de Brouwer AP, Morava E, Riemersma M, Schuurs-Hoeijmakers JH, Absmanner B, et al. Autosomal recessive dilated cardiomyopathy due to DOLK mutations results from abnormal dystroglycan O-mannosylation. PLoS genetics. 2011 Dec;7(12):e1002427. PubMed PMID: 22242004. Pubmed Central PMCID: 3248466. Epub 2012/01/14. eng.

238. Manya H, Chiba A, Yoshida A, Wang X, Chiba Y, Jigami Y, et al. Demonstration of mammalian protein O-mannosyltransferase activity: coexpression of POMT1 and POMT2 required for enzymatic activity. Proc Natl Acad Sci U S A. 2004 Jan 13;101(2):500-5. PubMed PMID: 14699049. Pubmed Central PMCID: 327176.

239. Akasaka-Manya K, Manya H, Nakajima A, Kawakita M, Endo T. Physical and functional association of human protein O-mannosyltransferases 1 and 2. J Biol Chem. 2006 Jul 14;281(28):19339-45. PubMed PMID: 16698797.

240. Akasaka-Manya K, Manya H, Hayashi M, Endo T. Different roles of the two components of human protein O-mannosyltransferase, POMT1 and POMT2. Biochem Biophys Res Commun. 2011 Aug 12;411(4):721-5. PubMed PMID: 21782786.

241. Hehr U, Uyanik G, Gross C, Walter MC, Bohring A, Cohen M, et al. Novel POMGnT1 mutations define broader phenotypic spectrum of muscle-eye-brain disease. Neurogenetics. 2007 Nov;8(4):279-88. PubMed PMID: 17906881.

242. Raducu M, Baets J, Fano O, Van Coster R, Cruces J. Promoter alteration causes transcriptional repression of the POMGNT1 gene in limb-girdle muscular dystrophy type 2O. Eur J Hum Genet. 2012 Sep;20(9):945-52. PubMed PMID: 22419172. Pubmed Central PMCID: 3421125.

243. Vuillaumier-Barrot S, Bouchet-Seraphin C, Chelbi M, Eude-Caye A, Charluteau E, Besson C, et al. Intragenic rearrangements in LARGE and POMGNT1 genes in severe dystroglycanopathies. Neuromuscul Disord. 2011 Nov;21(11):782-90. PubMed PMID: 21727005.

244. Balci B, Uyanik G, Dincer P, Gross C, Willer T, Talim B, et al. An autosomal recessive limb girdle muscular dystrophy (LGMD2) with mild mental retardation is allelic to Walker-Warburg syndrome (WWS) caused by a mutation in the POMT1 gene. Neuromuscul Disord. 2005 Apr;15(4):271-5. PubMed PMID: 15792865.

245. Bello L, Melacini P, Pezzani R, D'Amico A, Piva L, Leonardi E, et al. Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. Eur J Hum Genet. 2012 Dec;20(12):1234-9. PubMed PMID: 22549409. Pubmed Central PMCID: 3499746.

246. Yanagisawa A, Bouchet C, Quijano-Roy S, Vuillaumier-Barrot S, Clarke N, Odent S, et al. POMT2 intragenic deletions and splicing abnormalities causing congenital muscular dystrophy with mental retardation. Eur J Med Genet. 2009 Jul-Aug;52(4):201-6. PubMed PMID: 19138766.

247. Akasaka-Manya K, Manya H, Endo T. Mutations of the POMT1 gene found in patients with Walker-Warburg syndrome lead to a defect of protein O-mannosylation. Biochem Biophys Res Commun. 2004 Dec 3;325(1):75-9. PubMed PMID: 15522202.

248. Kanagawa M, Saito F, Kunz S, Yoshida-Moriguchi T, Barresi R, Kobayashi YM, et al. Molecular recognition by LARGE is essential for expression of functional dystroglycan. Cell. 2004 Jun 25;117(7):953-64. PubMed PMID: 15210115.

249. Inamori K, Yoshida-Moriguchi T, Hara Y, Anderson ME, Yu L, Campbell KP. Dystroglycan function requires xylosyl- and glucuronyltransferase activities of LARGE. Science. 2012 Jan 6;335(6064):93-6. PubMed PMID: 22223806. Epub 2012/01/10. eng.

250. Longman C, Brockington M, Torelli S, Jimenez-Mallebrera C, Kennedy C, Khalil N, et al. Mutations in the human LARGE gene cause MDC1D, a novel form of congenital muscular dystrophy with severe mental retardation and abnormal glycosylation of alpha-dystroglycan. Hum Mol Genet. 2003 Nov 1;12(21):2853-61. PubMed PMID: 12966029. Epub 2003/09/11. eng.

251. Brockington M, Torelli S, Prandini P, Boito C, Dolatshad NF, Longman C, et al. Localization and functional analysis of the LARGE family of glycosyltransferases: significance for muscular dystrophy. Hum Mol Genet. 2005 Mar 1;14(5):657-65. PubMed PMID: 15661757.

252. Bao X, Kobayashi M, Hatakeyama S, Angata K, Gullberg D, Nakayama J, et al. Tumor suppressor function of laminin-binding alpha-dystroglycan requires a distinct beta3-N-acetylglucosaminyltransferase. Proc Natl Acad Sci U S A. 2009 Jul 21;106(29):12109-14. PubMed PMID: 19587235. eng.

253. Lee PL, Kohler JJ, Pfeffer SR. Association of beta-1,3-N-acetylglucosaminyltransferase 1 and beta-1,4-galactosyltransferase 1, trans-Golgi enzymes involved in coupled poly-N-acetyllactosamine synthesis. Glycobiology. 2009 Jun;19(6):655-64. PubMed PMID: 19261593. Pubmed Central PMCID: 2682609.

254. Manzini MC, Tambunan DE, Hill RS, Yu TW, Maynard TM, Heinzen EL, et al. Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. Am J Hum Genet. 2012 Sep 7;91(3):541-7. PubMed PMID: 22958903. Epub 2012/09/11. eng.

255. Ogawa M, Nakamura N, Nakayama Y, Kurosaka A, Manya H, Kanagawa M, et al. GTDC2 modifies O-mannosylated alpha-dystroglycan in the endoplasmic reticulum to generate N-acetyl glucosamine epitopes reactive with CTD110.6 antibody. Biochem Biophys Res Commun. 2013 Oct 11;440(1):88-93. PubMed PMID: 24041696.

256. Jae LT, Raaben M, Riemersma M, van Beusekom E, Blomen VA, Velds A, et al. Deciphering the Glycosylome of Dystroglycanopathies Using Haploid Screens for Lassa Virus Entry. Science. 2013 Mar 21. PubMed PMID: 23519211. Epub 2013/03/23. Eng.

257. von Renesse A, Petkova MV, Lutzkendorf S, Heinemeyer J, Gill E, Hubner C, et al. POMK mutation in a family with congenital muscular dystrophy with merosin deficiency, hypomyelination, mild hearing deficit and intellectual disability. J Med Genet. 2014 Feb 20. PubMed PMID: 24556084.

258. Yoshida-Moriguchi T, Willer T, Anderson ME, Venzke D, Whyte T, Muntoni F, et al. SGK196 is a glycosylation-specific O-mannose kinase required for dystroglycan function. Science. 2013 Aug 23;341(6148):896-9. PubMed PMID: 23929950. Pubmed Central PMCID: 3848040.

259. Toda T, Kobayashi K, Kondo-Iida E, Sasaki J, Nakamura Y. The Fukuyama congenital muscular dystrophy story. Neuromuscul Disord. 2000 Mar;10(3):153-9. PubMed PMID: 10734260.

260. Kobayashi K, Nakahori Y, Miyake M, Matsumura K, Kondo-Iida E, Nomura Y, et al. An ancient retrotransposal insertion causes Fukuyama-type congenital muscular dystrophy. Nature. 1998 Jul 23;394(6691):388-92. PubMed PMID: 9690476. Epub 1998/08/05. eng.

261. Watanabe M, Kobayashi K, Jin F, Park KS, Yamada T, Tokunaga K, et al. Founder SVA retrotransposal insertion in Fukuyama-type congenital muscular dystrophy and its origin in Japanese and Northeast Asian populations. Am J Med Genet A. 2005 Nov 1;138(4):344-8. PubMed PMID: 16222679.

262. Taniguchi-Ikeda M, Kobayashi K, Kanagawa M, Yu CC, Mori K, Oda T, et al. Pathogenic exon-trapping by SVA retrotransposon and rescue in Fukuyama muscular dystrophy. Nature. 2011 Oct 6;478(7367):127-31. PubMed PMID: 21979053. Pubmed Central PMCID: 3412178.

263. Kondo-Iida E, Kobayashi K, Watanabe M, Sasaki J, Kumagai T, Koide H, et al. Novel mutations and genotype-phenotype relationships in 107 families with Fukuyama-type congenital muscular dystrophy (FCMD). Hum Mol Genet. 1999 Nov;8(12):2303-9. PubMed PMID: 10545611.

264. Silan F, Yoshioka M, Kobayashi K, Simsek E, Tunc M, Alper M, et al. A new mutation of the fukutin gene in a non-Japanese patient. Ann Neurol. 2003 Mar;53(3):392-6. PubMed PMID: 12601708.

265. Cotarelo RP, Valero MC, Prados B, Pena A, Rodriguez L, Fano O, et al. Two new patients bearing mutations in the fukutin gene confirm the relevance of this gene in Walker-Warburg syndrome. Clin Genet. 2008 Feb;73(2):139-45. PubMed PMID: 18177472.

266. Godfrey C, Escolar D, Brockington M, Clement EM, Mein R, Jimenez-Mallebrera C, et al. Fukutin gene mutations in steroid-responsive limb girdle muscular dystrophy. Ann Neurol. 2006 Nov;60(5):603-10. PubMed PMID: 17044012.

267. Yis U, Uyanik G, Heck PB, Smitka M, Nobel H, Ebinger F, et al. Fukutin mutations in non-Japanese patients with congenital muscular dystrophy: less severe mutations predominate in patients with a non-Walker-Warburg phenotype. Neuromuscul Disord. 2011 Jan;21(1):20-30. PubMed PMID: 20961758.

268. Hayashi YK, Ogawa M, Tagawa K, Noguchi S, Ishihara T, Nonaka I, et al. Selective deficiency of alpha-dystroglycan in Fukuyama-type congenital muscular dystrophy. Neurology. 2001 Jul 10;57(1):115-21. PubMed PMID: 11445638.

269. Brockington M, Blake DJ, Prandini P, Brown SC, Torelli S, Benson MA, et al. Mutations in the fukutin-related protein gene (FKRP) cause a form of congenital muscular dystrophy with secondary laminin alpha2 deficiency and abnormal glycosylation of alpha-dystroglycan. Am J Hum Genet. 2001 Dec;69(6):1198-209. PubMed PMID: 11592034. Pubmed Central PMCID: 1235559. Epub 2001/10/10. eng.

270. Brockington M, Yuva Y, Prandini P, Brown SC, Torelli S, Benson MA, et al. Mutations in the fukutin-related protein gene (FKRP) identify limb girdle muscular dystrophy 2I as a milder allelic variant of congenital muscular dystrophy MDC1C. Hum Mol Genet. 2001 Dec 1;10(25):2851-9. PubMed PMID: 11741828.

271. Mercuri E, Brockington M, Straub V, Quijano-Roy S, Yuva Y, Herrmann R, et al. Phenotypic spectrum associated with mutations in the fukutin-related protein gene. Ann Neurol. 2003 Apr;53(4):537-42. PubMed PMID: 12666124.

272. Bourteel H, Vermersch P, Cuisset JM, Maurage CA, Laforet P, Richard P, et al. Clinical and mutational spectrum of limb-girdle muscular dystrophy type 2I in 11 French patients. J Neurol Neurosurg Psychiatry. 2009 Dec;80(12):1405-8. PubMed PMID: 19917824.

273. Saito Y, Mizuguchi M, Oka A, Takashima S. Fukutin protein is expressed in neurons of the normal developing human brain but is reduced in Fukuyama-type congenital muscular dystrophy brain. Ann Neurol. 2000 Jun;47(6):756-64. PubMed PMID: 10852541.

274. Yamamoto T, Kato Y, Karita M, Takeiri H, Muramatsu F, Kobayashi M, et al. Fukutin expression in glial cells and neurons: implication in the brain lesions of Fukuyama congenital muscular dystrophy. Acta Neuropathol. 2002 Sep;104(3):217-24. PubMed PMID: 12172906.

275. Matsumoto H, Noguchi S, Sugie K, Ogawa M, Murayama K, Hayashi YK, et al. Subcellular localization of fukutin and fukutin-related protein in muscle cells. J Biochem. 2004 Jun;135(6):709-12. PubMed PMID: 15213246.

276. Tachikawa M, Kanagawa M, Yu CC, Kobayashi K, Toda T. Mislocalization of fukutin protein by disease-causing missense mutations can be rescued with treatments directed at folding amelioration. J Biol Chem. 2012 Mar 9;287(11):8398-406. PubMed PMID: 22275357. Pubmed Central PMCID: 3318729.

277. Xiong H, Kobayashi K, Tachikawa M, Manya H, Takeda S, Chiyonobu T, et al. Molecular interaction between fukutin and POMGnT1 in the glycosylation pathway of alpha-dystroglycan. Biochem Biophys Res Commun. 2006 Dec 1;350(4):935-41. PubMed PMID: 17034757.

278. Beedle AM, Nienaber PM, Campbell KP. Fukutin-related protein associates with the sarcolemmal dystrophin-glycoprotein complex. J Biol Chem. 2007 Jun 8;282(23):16713-7. PubMed PMID: 17452335.

279. Esapa CT, McIlhinney RA, Blake DJ. Fukutin-related protein mutations that cause congenital muscular dystrophy result in ER-retention of the mutant protein in cultured cells. Hum Mol Genet. 2005 Jan 15;14(2):295-305. PubMed PMID: 15574464.

280. Dolatshad NF, Brockington M, Torelli S, Skordis L, Wever U, Wells DJ, et al. Mutated fukutin-related protein (FKRP) localises as wild type in differentiated muscle cells. Exp Cell Res. 2005 Oct 1;309(2):370-8. PubMed PMID: 16055117.

281. Willer T, Lee H, Lommel M, Yoshida-Moriguchi T, de Bernabe DB, Venzke D, et al. ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nat Genet. 2012;44(5):575-80. PubMed PMID: 22522420. Pubmed Central PMCID: 3371168. Epub 2012/04/24. eng.

282. Roscioli T, Kamsteeg EJ, Buysse K, Maystadt I, van Reeuwijk J, van den Elzen C, et al. Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of alpha-dystroglycan. Nat Genet. 2012;44(5):581-5. PubMed PMID: 22522421. Epub 2012/04/24. eng.

283. Cirak S, Foley AR, Herrmann R, Willer T, Yau S, Stevens E, et al. ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. Brain : a journal of neurology. 2013 Jan 3. PubMed PMID: 23288328. Epub 2013/01/05. Eng.

284. Vuillaumier-Barrot S, Bouchet-Seraphin C, Chelbi M, Devisme L, Quentin S, Gazal S, et al. Identification of Mutations in TMEM5 and ISPD as a Cause of Severe Cobblestone Lissencephaly. Am J Hum Genet. 2012 Dec 7;91(6):1135-43. PubMed PMID: 23217329. Pubmed Central PMCID: 3516603. Epub 2012/12/12. eng.

285. Peat RA, Smith JM, Compton AG, Baker NL, Pace RA, Burkin DJ, et al. Diagnosis and etiology of congenital muscular dystrophy. Neurology. 2008 Jul 29;71(5):312-21. PubMed PMID: 18160674.

286. Messina S, Mora M, Pegoraro E, Pini A, Mongini T, D'Amico A, et al. POMT1 and POMT2 mutations in CMD patients: a multicentric Italian study. Neuromuscul Disord. 2008 Jul;18(7):565-71. PubMed PMID: 18513969.

287. Mercuri E, Messina S, Bruno C, Mora M, Pegoraro E, Comi GP, et al. Congenital muscular dystrophies with defective glycosylation of dystroglycan: a population study. Neurology. 2009 May 26;72(21):1802-9. PubMed PMID: 19299310. eng.

288. Vervoort VS, Holden KR, Ukadike KC, Collins JS, Saul RA, Srivastava AK. POMGnT1 gene alterations in a family with neurological abnormalities. Ann Neurol. 2004 Jul;56(1):143-8. PubMed PMID: 15236414.

289. Yanagisawa A, Bouchet C, Van den Bergh PY, Cuisset JM, Viollet L, Leturcq F, et al. New POMT2 mutations causing congenital muscular dystrophy: identification of a founder mutation. Neurology. 2007 Sep 18;69(12):1254-60. PubMed PMID: 17634419.

290. Clement E, Mercuri E, Godfrey C, Smith J, Robb S, Kinali M, et al. Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. Ann Neurol. 2008 Nov;64(5):573-82. PubMed PMID: 19067344.

291. Van Reeuwijk J, Olderode-Berends MJ, Van den Elzen C, Brouwer OF, Roscioli T, Van Pampus MG, et al. A homozygous FKRP start codon mutation is associated with Walker-Warburg syndrome, the severe end of the clinical spectrum. Clin Genet. 2010 Sep;78(3):275-81. PubMed PMID: 20236121.

292. Jimenez-Mallebrera C, Torelli S, Feng L, Kim J, Godfrey C, Clement E, et al. A comparative study of alpha-dystroglycan glycosylation in dystroglycanopathies suggests that the hypoglycosylation of alpha-dystroglycan does not consistently correlate with clinical severity. Brain Pathol. 2009 Oct;19(4):596-611. PubMed PMID: 18691338. Pubmed Central PMCID: 2860390. Epub 2008/08/12. eng.

293. Saredi S, Ardissone A, Ruggieri A, Mottarelli E, Farina L, Rinaldi R, et al. Novel POMGNT1 point mutations and intragenic rearrangements associated with muscle-eye-brain disease. J Neurol Sci. 2012 Jul 15;318(1-2):45-50. PubMed PMID: 22554691. Pubmed Central PMCID: 3405532.

294. Howe K, Clark MD, Torroja CF, Torrance J, Berthelot C, Muffato M, et al. The zebrafish reference genome sequence and its relationship to the human genome. Nature. 2013 Apr 25;496(7446):498-503. PubMed PMID: 23594743. Pubmed Central PMCID: 3703927.

295. Lin YY. Muscle diseases in the zebrafish. Neuromuscular disorders : NMD. 2012 May 28. PubMed PMID: 22647769. Epub 2012/06/01. Eng.

296. Parsons MJ, Campos I, Hirst EM, Stemple DL. Removal of dystroglycan causes severe muscular dystrophy in zebrafish embryos. Development. 2002 Jul;129(14):3505-12. PubMed PMID: 12091319. Epub 2002/07/02. eng.

297. Garrity DM, Childs S, Fishman MC. The heartstrings mutation in zebrafish causes heart/fin Tbx5 deficiency syndrome. Development. 2002 Oct;129(19):4635-45. PubMed PMID: 12223419.

298. Gerlai R. Social behavior of zebrafish: From synthetic images to biological mechanisms of shoaling. J Neurosci Methods. 2014 May 2. PubMed PMID: 24793400.

299. Kettleborough RN, Busch-Nentwich EM, Harvey SA, Dooley CM, de Bruijn E, van Eeden F, et al. A systematic genome-wide analysis of zebrafish protein-coding gene function. Nature. 2013 Apr 25;496(7446):494-7. PubMed PMID: 23594742. Pubmed Central PMCID: 3743023.

300. Blackburn PR, Campbell JM, Clark KJ, Ekker SC. The CRISPR system--keeping zebrafish gene targeting fresh. Zebrafish. 2013 Mar;10(1):116-8. PubMed PMID: 23536990. Pubmed Central PMCID: 3629780.

301. Fu Y, Foden JA, Khayter C, Maeder ML, Reyon D, Joung JK, et al. High-frequency off-target mutagenesis induced by CRISPR-Cas nucleases in human cells. Nat Biotechnol. 2013 Sep;31(9):822-6. PubMed PMID: 23792628. Pubmed Central PMCID: 3773023.

302. Robu ME, Larson JD, Nasevicius A, Beiraghi S, Brenner C, Farber SA, et al. p53 activation by knockdown technologies. PLoS genetics. 2007 May 25;3(5):e78. PubMed PMID: 17530925. eng.

303. Gerety SS, Wilkinson DG. Morpholino artifacts provide pitfalls and reveal a novel role for pro-apoptotic genes in hindbrain boundary development. Dev Biol. 2011 Feb 15;350(2):279-89. PubMed PMID: 21145318. Pubmed Central PMCID: 3111810. Epub 2010/12/15. eng.

304. Moore CJ, Goh HT, Hewitt JE. Genes required for functional glycosylation of dystroglycan are conserved in zebrafish. Genomics. 2008 Sep;92(3):159-67. PubMed PMID: 18632251. Epub 2008/07/18. eng.

305. Kawahara G, Guyon JR, Nakamura Y, Kunkel LM. Zebrafish models for human FKRP muscular dystrophies. Hum Mol Genet. 2010 Feb 15;19(4):623-33. PubMed PMID: 19955119. Pubmed Central PMCID: 2807370. Epub 2009/12/04. eng.

306. Avsar-Ban E, Ishikawa H, Manya H, Watanabe M, Akiyama S, Miyake H, et al. Protein O-mannosylation is necessary for normal embryonic development in zebrafish. Glycobiology. 2010 Sep;20(9):1089-102. PubMed PMID: 20466645. Epub 2010/05/15. eng.

307. Gupta V, Kawahara G, Gundry SR, Chen AT, Lencer WI, Zhou Y, et al. The zebrafish dag1 mutant: a novel genetic model for dystroglycanopathies. Hum Mol Genet. 2011 May 1;20(9):1712-25. PubMed PMID: 21296866. Pubmed Central PMCID: 3071669.

308. Chiyonobu T, Sasaki J, Nagai Y, Takeda S, Funakoshi H, Nakamura T, et al. Effects of fukutin deficiency in the developing mouse brain. Neuromuscul Disord. 2005 Jun;15(6):416-26. PubMed PMID: 15907289.

309. Stalnaker SH, Aoki K, Lim JM, Porterfield M, Liu M, Satz JS, et al. Glycomic analyses of mouse models of congenital muscular dystrophy. J Biol Chem. 2011 Jun 17;286(24):21180-90. PubMed PMID: 21460210. Pubmed Central PMCID: 3122180.

310. Lin YY, White RJ, Torelli S, Cirak S, Muntoni F, Stemple DL. Zebrafish Fukutin family proteins link the unfolded protein response with dystroglycanopathies. Hum Mol Genet. 2011 May 1;20(9):1763-75. PubMed PMID: 21317159. eng.

311. Barresi R, Michele DE, Kanagawa M, Harper HA, Dovico SA, Satz JS, et al. LARGE can functionally bypass alpha-dystroglycan glycosylation defects in distinct congenital muscular dystrophies. Nat Med. 2004 Jul;10(7):696-703. PubMed PMID: 15184894. Epub 2004/06/09. eng.

312. Yu M, He Y, Wang K, Zhang P, Zhang S, Hu H. Adeno-associated viral-mediated LARGE gene therapy rescues the muscular dystrophic phenotype in mouse models of dystroglycanopathy. Hum Gene Ther. 2013 Mar;24(3):317-30. PubMed PMID: 23379513. Pubmed Central PMCID: 3609641.

313. Carss KJ, Stevens E, Foley AR, Cirak S, Riemersma M, Torelli S, et al. Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of alpha-dystroglycan. Am J Hum Genet. 2013 Jul 11;93(1):29-41. PubMed PMID: 23768512. Pubmed Central PMCID: 3710768.

314. Stevens E, Carss KJ, Cirak S, Foley AR, Torelli S, Willer T, et al. Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of alpha-Dystroglycan. Am J Hum Genet. 2013 Mar 7;92(3):354-65. PubMed PMID: 23453667. Pubmed Central PMCID: 3591840. Epub 2013/03/05. eng.

315. Hiruma T, Togayachi A, Okamura K, Sato T, Kikuchi N, Kwon YD, et al. A novel human beta1,3-N-acetylgalactosaminyltransferase that synthesizes a unique carbohydrate structure, GalNAcbeta1-3GlcNAc. J Biol Chem. 2004 Apr 2;279(14):14087-95. PubMed PMID: 14724282. eng.

316. Harrison R, Hitchen PG, Panico M, Morris HR, Mekhaiel D, Pleass RJ, et al. Glycoproteomic characterization of recombinant mouse alpha-dystroglycan. Glycobiology. 2012 May;22(5):662-75. PubMed PMID: 22241827. Pubmed Central PMCID: 3311285.

317. Ning B, Elbein AD. Cloning, expression and characterization of the pig liver GDP-mannose pyrophosphorylase. Evidence that GDP-mannose and GDP-Glc pyrophosphorylases are different proteins. Eur J Biochem. 2000 Dec;267(23):6866-74. PubMed PMID: 11082198. eng.

318. Malicki J, Jo H, Wei X, Hsiung M, Pujic Z. Analysis of gene function in the zebrafish retina. Methods. 2002 Dec;28(4):427-38. PubMed PMID: 12507461. Epub 2003/01/01. eng.

319. Abramoff MD, Magalhaes, P.J., Ram, S.J. Image Processing with ImageJ. Biophotonics International. 2004;11(7):36-42.

320. Thisse B, Thisse, C. Fast Release Clones: A High Throughput Expression Analysis. ZFIN Direct Data Submission 2004. Available from: http://zfin.org.

321. Thisse B, Pflumio, S., Fürthauer, M., Loppin, B., Heyer, V., Degrave, A., Woehl, R., Lux, A., Steffan, T., Charbonnier, X.Q., Thisse, C. Expression of the zebrafish genome during embryogenesis (NIH R01 RR15402). ZFIN Direct Data Submission (http://zfinorg) 2001.

322. Bassett DI, Bryson-Richardson RJ, Daggett DF, Gautier P, Keenan DG, Currie PD. Dystrophin is required for the formation of stable muscle attachments in the zebrafish embryo. Development. 2003 Dec;130(23):5851-60. PubMed PMID: 14573513. eng.

323. Niederriter AR, Davis EE, Golzio C, Oh EC, Tsai IC, Katsanis N. In vivo modeling of the morbid human genome using Danio rerio. Journal of visualized experiments : JoVE. 2013 (78):e50338. PubMed PMID: 23995499. Pubmed Central PMCID: 3856313.

324. Golzio C, Willer J, Talkowski ME, Oh EC, Taniguchi Y, Jacquemont S, et al. KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature. 2012 May 17;485(7398):363-7. PubMed PMID: 22596160. Pubmed Central PMCID: 3366115. Epub 2012/05/19. eng.

325. Piepenburg O, Grimmer D, Williams PH, Smith JC. Activin redux: specification of mesodermal pattern in Xenopus by graded concentrations of endogenous activin B. Development. 2004 Oct;131(20):4977-86. PubMed PMID: 15371302.

326. Eisen JS, Smith JC. Controlling morpholino experiments: don't stop making antisense. Development. 2008 May;135(10):1735-43. PubMed PMID: 18403413.

327. Lisenbee CS, Karnik SK, Trelease RN. Overexpression and mislocalization of a tail-anchored GFP redefines the identity of peroxisomal ER. Traffic. 2003 Jul;4(7):491-501. PubMed PMID: 12795694.

328. Hedberg C, Oldfors A, Darin N. B3GALNT2 is a gene associated with congenital muscular dystrophy with brain malformations. Eur J Hum Genet. 2014 May;22(5):707-10. PubMed PMID: 24084573. Pubmed Central PMCID: 3992579.

329. Cao W, Henry MD, Borrow P, Yamada H, Elder JH, Ravkov EV, et al. Identification of alpha-dystroglycan as a receptor for lymphocytic choriomeningitis virus and Lassa fever virus. Science. 1998 Dec 11;282(5396):2079-81. PubMed PMID: 9851928.

330. Raphael AR, Couthouis J, Sakamuri S, Siskind C, Vogel H, Day JW, et al. Congenital muscular dystrophy and generalized epilepsy caused by GMPPB mutations. Brain Res. 2014 Apr 26. PubMed PMID: 24780531.

331. Sharma V, Ichikawa M, He P, Scott DA, Bravo Y, Dahl R, et al. Phosphomannose isomerase inhibitors improve N-glycosylation in selected phosphomannomutase-deficient fibroblasts. J Biol Chem. 2011 Nov 11;286(45):39431-8. PubMed PMID: 21949237. Pubmed Central PMCID: 3234766. Epub 2011/09/29. eng.

332. Koehler K, Malik M, Mahmood S, Giesselmann S, Beetz C, Hennings JC, et al. Mutations in GMPPA cause a glycosylation disorder characterized by intellectual disability and autonomic dysfunction. Am J Hum Genet. 2013 Oct 3;93(4):727-34. PubMed PMID: 24035193. Pubmed Central PMCID: 3791256.

333. Davis AJ, Perugini MA, Smith BJ, Stewart JD, Ilg T, Hodder AN, et al. Properties of GDP-mannose pyrophosphorylase, a critical enzyme and drug target in Leishmania mexicana. J Biol Chem. 2004 Mar 26;279(13):12462-8. PubMed PMID: 14718535. Epub 2004/01/14. eng.

334. Warit S, Zhang N, Short A, Walmsley RM, Oliver SG, Stateva LI. Glycosylation deficiency phenotypes resulting from depletion of GDP-mannose pyrophosphorylase in two yeast species. Mol Microbiol. 2000 Jun;36(5):1156-66. PubMed PMID: 10844699. Epub 2000/06/09. eng.

335. Zhang N, Gardner DC, Oliver SG, Stateva LI. Down-regulation of the expression of PKC1 and SRB1/PSA1/VIG9, two genes involved in cell wall integrity in Saccharomyces cerevisiae, causes flocculation. Microbiology. 1999 Feb;145 (Pt 2):309-16. PubMed PMID: 10075413. Epub 1999/03/13. eng.

336. Jiang H, Ouyang H, Zhou H, Jin C. GDP-mannose pyrophosphorylase is essential for cell wall integrity, morphogenesis and viability of Aspergillus fumigatus. Microbiology. 2008 Sep;154(Pt 9):2730-9. PubMed PMID: 18757806. Epub 2008/09/02. eng.

337. Qin C, Qian W, Wang W, Wu Y, Yu C, Jiang X, et al. GDP-mannose pyrophosphorylase is a genetic determinant of ammonium sensitivity in Arabidopsis thaliana. Proc Natl Acad Sci U S A. 2008 Nov 25;105(47):18308-13. PubMed PMID: 19011088. Pubmed Central PMCID: 2587558. Epub 2008/11/18. eng.

338. Keller R, Renz FS, Kossmann J. Antisense inhibition of the GDP-mannose pyrophosphorylase reduces the ascorbate content in transgenic plants leading to developmental changes during senescence. Plant J. 1999 Jul;19(2):131-41. PubMed PMID: 10476060. Epub 1999/09/04. eng.

339. Denton H, Fyffe S, Smith TK. GDP-mannose pyrophosphorylase is essential in the bloodstream form of Trypanosoma brucei. Biochem J. 2010 Feb 1;425(3):603-14. PubMed PMID: 19919534. Epub 2009/11/19. eng.

340. Fu Y, Sander JD, Reyon D, Cascio VM, Joung JK. Improving CRISPR-Cas nuclease specificity using truncated guide RNAs. Nat Biotechnol. 2014 Mar;32(3):279-84. PubMed PMID: 24463574. Pubmed Central PMCID: 3988262.

341. Barrangou R, Fremaux C, Deveau H, Richards M, Boyaval P, Moineau S, et al. CRISPR provides acquired resistance against viruses in prokaryotes. Science. 2007 Mar 23;315(5819):1709-12. PubMed PMID: 17379808.

342. Barrangou R. RNA-mediated programmable DNA cleavage. Nat Biotechnol. 2012 Sep;30(9):836-8. PubMed PMID: 22965054.

343. Faridmoayer A, Fentabil MA, Haurat MF, Yi W, Woodward R, Wang PG, et al. Extreme substrate promiscuity of the Neisseria oligosaccharyl transferase involved in protein O-glycosylation. J Biol Chem. 2008 Dec 12;283(50):34596-604. PubMed PMID: 18930921. Pubmed Central PMCID: 3259870.

344. Gantt RW, Goff RD, Williams GJ, Thorson JS. Probing the aglycon promiscuity of an engineered glycosyltransferase. Angew Chem Int Ed Engl. 2008;47(46):8889-92. PubMed PMID: 18924204. Pubmed Central PMCID: 2963038.

345. Gantt RW, Peltier-Pain P, Singh S, Zhou M, Thorson JS. Broadening the scope of glycosyltransferase-catalyzed sugar nucleotide synthesis. Proc Natl Acad Sci U S A. 2013 May 7;110(19):7648-53. PubMed PMID: 23610417. Pubmed Central PMCID: 3651490.

346. Vuillaumier-Barrot S, Le Bizec C, de Lonlay P, Barnier A, Mitchell G, Pelletier V, et al. Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type Ib. J Med Genet. 2002 Nov;39(11):849-51. PubMed PMID: 12414827. Pubmed Central PMCID: 1735008.

347. Niehues R, Hasilik M, Alton G, Korner C, Schiebe-Sukumar M, Koch HG, et al. Carbohydrate-deficient glycoprotein syndrome type Ib. Phosphomannose isomerase

deficiency and mannose therapy. J Clin Invest. 1998 Apr 1;101(7):1414-20. PubMed PMID: 9525984. Pubmed Central PMCID: 508719.

348. Chu J, Mir A, Gao N, Rosa S, Monson C, Sharma V, et al. A zebrafish model of congenital disorders of glycosylation with phosphomannose isomerase deficiency reveals an early opportunity for corrective mannose supplementation. Dis Model Mech. 2012 Sep 6. PubMed PMID: 22899857. Epub 2012/08/18. Eng.