

Automated Splice Site Analysis Server citations:

1. Leman, AR, Pearce, DA, Rothberg, PG. Human Gene Mutation. Gene symbol: *CLN3*. Disease: Juvenile Neuronal Ceroid Lipofuscinosis (Batten disease). *Human Genetics* 116: 236, 2005.
2. V Wessagowit, S-C Kim, S W Oh and JA McGrath. Genotype Phenotype Correlation in Recessive Dystrophic Epidermolysis Bullosa: When Missense Doesn't Make Sense. *Journal of Investigative Dermatology*, 124: 863-866, 2005.
3. A Gaedigk, L Ndjountche, JS Leeder and LD Bradford. Limited association of the 2988G>A single nucleotide polymorphism with *CYP2D6*41* in black subjects. *Clinical Pharmacology & Therapeutics* 77: 228-230, 2005.
4. A Henriksen, T Zeynep, N Tommerup, L Tranebj, LA Larsen. Identification of a Novel *EYAI* Splice-Site Mutation in a Danish Branchio-Oto-Renal Syndrome Family. *Genetic Testing* 8(4): 404-406, 2004.
5. T Fasano, L Bocchi, L Pisciotta, S Bertolini and S Calandra, Denaturing high-performance liquid chromatography in the detection of *ABCA1* gene mutations in familial HDL deficiency. *Journal of Lipid Research* 46: 817-822, 2005.
6. E Di Leo , F Panico , P Tarugi , C Battisti , A Federico , S Calandra, A point mutation in the lariat branch point of intron 6 of *NPC1* as the cause of abnormal pre-mRNA splicing in Niemann-Pick type C disease . *Human Mutation, Mutation in Brief #758 (2004) Online*
7. A Gaedigk, A Bhatena, L Ndjountche, RE Pearce, SM Abdel-Rahman, SW Alander, LD Bradford, PK Rogan and JS Leeder, Identification and characterization of novel sequence variations in the cytochrome P450D6 (*CYP2D6*) gene in African Americans. *The Pharmacogenomics Journal* 5: 173-182, 2005.
8. Nalla V and Rogan PK. Automated splice site mutation analysis by information theory (<https://splice.uwo.ca>). *Hum Mut.* 25:334-342, 2005.
9. Nalla V. Automated splice site analysis (<https://splice.uwo.ca>; Dissertation). University of Missouri-Kansas City, 2005.
10. Hageman G et al. A common haplotype in the complement regulatory gene factor H (*HF1/CFH*) predisposes individuals to age-related macular degeneration. *Proc Natl. Acad. Sci.* 102(20):7227-7232, 2005.
11. WS Oetting and T Tabone, The 2004 Human Genome Variation Society scientific meeting . *Human Mutation* 26:160-163, 2005.
12. SB Koukouritaki, MT Poch, ET Cabacungan, DG McCarver, and RN Hines, Discovery of Novel Flavin-Containing Monooxygenase 3 (*FMO3*) Single Nucleotide Polymorphisms and Functional Analysis of Upstream Haplotype Variants . *Molecular Pharmacology* 68:383-392, 2005.
13. V Wessagowit, VK Nalla, PK Rogan, JA McGrath. Normal and abnormal mechanisms of gene splicing and relevance to inherited skin diseases. *Journal of Dermatological Science*, 40(2):73-84, 2005.
14. The American College of Medical Genetics Laboratory Quality Assurance Committee, "[Technical Standards and Guidelines:Molecular Genetic Testing for Rare Disorders,](#)" [Paragraph: URD 7.4.1.3](#), 2005.
15. M Fornage, CR Lee, PA Doris, MS Bray, G Heiss, DC Zeldin and E Boerwinkle. The soluble epoxide hydrolase gene harbors sequence variation associated with susceptibility

- to and protection from incident ischemic stroke *Human Molecular Genetics*, 14(19): 2829-2837, 2005.
16. G Hobson, Z Huang, K Sperle, E Sistermans, PK Rogan, JY Garbern, E Kolodny, S Naidu, and F Cambi. Splice site contribution in alternative splicing of *PLP* and *DM20*: molecular studies in oligodendrocytes. *Human Mutation*, DOI: 10.1002/humu.20276.
 17. S Lancellotti, E Di Leo, S Calandra, and P Tarugi, DIFETTO DI SPLICING DEL PRE-mRNA DELL'APOLIPOPROTEINA B NEL FEGATO DI PAZIENTI CON IPOBETALIPOPROTEINEMIA FAMILIARE. *Patologia genetica*, 2005 (www.sip2005.unimore.it/genetica.pdf)
 18. Wessagowit V and JA McGrath. Clinical and Molecular Significance of Splice Site Mutations in the Plakophilin 1 Gene in Patients with Ectodermal Dysplasia-Skin Fragility Syndrome. *Acta Dermato-Venereologica*, 85(5): 386-388, 2005.
 19. Yu H, SB Patel. Recent insights into the Smith-Lemli-Opitz syndrome. *Clin Genet*, 68: 383-391, 2005.
 20. Zhang Q, F Zulfiqar, SA Riazuddin, X Xiao, A Yasmeen, PK Rogan, R Caruso, PA Sieving, S Riazuddin, JF Hejtmancik. A variant form of Oguchi disease mapped to 13q34 associated with partial deletion of *GRK1* gene. *Mol Vision*, 11: 977-985, 2005.
 21. Maddalena A, S Bale, S Das, W Grody, SP Richards. Technical Standards and Guidelines: Molecular Genetic Testing for Ultra-Rare Disorders. *Genetics in Medicine*, 7(8): 571-583, 2005.
 22. H Russcher, P Smit, E van Rossum, E van den Akker, A Brinkmann, L de Heide, F de Jong, J Koper, and S Lamberts. Strategies for the Characterisation of Disorders in Cortisol Sensitivity. *J Clin Endocrin Metab.*, doi:10.1210/jc.2005-2212, 91[2]: 694-701, 2006.
 23. A Tartaglia-Polcini, C Bonnard, A Micheloni, F Cianfarani, A Andre, G Zambruno, A Hovnanian and M D'Alessio. *SPINK5*, the defective gene in Netherton Syndrome, encodes multiple *LEKTI* isoforms derived from alternative pre-mRNA processing. *J. Invest. Derm.*, doi:10:1038/sj.jid.5700015, 2005.
 24. Parslow GR. Multimedia of Biochemistry and Molecular Biology Education: Websites of Note. *Biochem. Educ.*, 34(1): 55-57, 2006.
 25. Skipper L, Shen H, Chua E, Bonnard C, Kolatkar P, Tan LC, Jamora RD, Puvan K, Puong KY, Zhao Y, Pavanni R, Wong MC, Yuen Y, Farrer M, Liu JJ, Tan EK. Analysis of *LRRK2* functional domains in nondominant Parkinson disease. *Neurology*, 65(8):1319-21, 2005.
 26. Cox DG, JBA Crusius, PHM Peeters, HB Bueno-de-Mesquita, AS Pe, F Canzian. A haplotype of prostaglandin synthase 2/cyclooxygenase 2 is involved in the susceptibility to inflammatory bowel disease. *World J Gastroenterol*, 11(38):6003-6008, 2005.
 27. Tazi J, Durand S, and P Jeanteur. The spliceosome: a novel multi-faceted target for therapy. *Trends in Biochem. Sciences*, 30(8):469-478, 2005.
 28. Titeux M, Mejí JE, Mejlumian L, Bourthoumieu S, Mirval S, Tonasso L, Heller M, Prost-Squarcioni C, Hovnanian A. Recessive dystrophic epidermolysis bullosa caused by *COL7A1* hemizyosity and a missense mutation with complex effects on splicing. *Human Mutation*, 27:291-292, 2006.
 29. Wang P, Guo X, Jia X, Li S, Xiao X, Zhang Q. Novel mutations of the *PAX6* gene identified in Chinese patients with aniridia. *Molecular Vision* 12:644-8, 2006.

30. Cambi F, Hobson G, Sperle K, Huang Z, Garbern J, and Rogan PK. Is PLP necessary for brain development and function? "The human perspective." Workshop W01: PLP and PLP Mutants – What's New, *J. Neurochemistry* 96(s1): 12-13, 2006.
31. Russcher H. Glucocorticoid Receptor Variants Modulate the Sensitivity to Cortisol (Dissertation), Erasmus University, Rotterdam, Netherlands, 2006.
32. Pasmooij AMG. Revertant mosaicism in epidermolysis bullosa due to different second site mutations in LAMB3 (Dissertation, Chapter 3), University Medical Center Groningen, University of Groningen, Netherlands, 2006.
33. Mukhopadhyay A, Nikopoulos K, Maugeri A, de Brouwer AP, van Nouhuys CE, Boon CJ, Perveen R, Zegers HA, Wittebol-Post D, van den Biesen PR, van der Velde-Visser SD, Brunner HG, Black GC, Hoyng CB, Cremers FP. Erosive vitreoretinopathy and wagner disease are caused by intronic mutations in *CSPG2*/Versican that result in an imbalance of splice variants. *Invest Ophthalmol Vis Sci.* 47(8):3565-72, 2006.
34. Zaffanello M, Taranta A, Palma A, Bettinelli A, Marseglia GL, Emma F. Type IV Bartter syndrome: report of two new cases. *Pediatr Nephrol.* 21(6):766-70, 2006.
35. Tartaglia-Polcini A, Bonnart C, Micheloni A, Cianfarani F, Andre A, Zambruno G, Hovnanian A, D'Alessio M. *SPINK5*, the defective gene in netherton syndrome, encodes multiple LEKTI isoforms derived from alternative pre-mRNA processing. *J Invest Dermatol.* 126(2):315-24, 2006.
36. Hube F, Guo J, Chooniedass-Kothari S, Cooper C, Hamedani MK, Dibrov AA, Blanchard AA, Wang X, Deng G, Myal Y, Leygue E. Alternative splicing of the first intron of the steroid receptor RNA activator (SRA) participates in the generation of coding and noncoding RNA isoforms in breast cancer cell lines. *DNA Cell Biol.* 25(7):418-28, 2006.
37. Buratti E, Baralle M, Baralle FE. Defective splicing, disease and therapy: searching for master checkpoints in exon definition. *Nucleic Acids Res.* 34(12):3494-510, 2006.
38. Day INM. *IDDM2* locus: 5' noncoding intron I splicing and translational efficiency effects of *INS* -23HphI – more than a tag for the *INS* promoter *VNTR*. Hum Gen Var Society, 2006 (abstracts/hgvs.org/Helsenki/Presentations/Day.ppt).
39. Hiller M, Huse K, Szafranskzi K, Rosenstiel P, Schreiber S, Backofen R, Platzer M. Phylogenetically widespread alternative splicing at unusual GYNGYN donors. *Genome Biol.* 7(7):R65, 2006
40. Tosetto E, Ghiggeri GM, Emma F, Barbano G, Carrea A, Vezzoli G, Torregrossa R, Cara M, Ripanti G, Ammenti A, Peruzzi L, Murer L, Ratsch IM, Citron L, Gambaro G, D'angelo A, Anglani F. Phenotypic and genetic heterogeneity in Dent's disease--the results of an Italian collaborative study. *Nephrol Dial Transplant.* 21(9):2452-63, 2006.
41. Gaedigk A, Baker DW, Totah RA, Gaedigk R, Pearce RE, Vyhlidal CA, Zeldin DC, Leeder JS. Variability of *CYP2J2* Expression in Human Fetal Tissues. *J Pharmacol Exp Ther.* 319(2):523-32, 2006.
42. Vorechovsky I. Aberrant 3' splice sites in human disease genes: mutation pattern, nucleotide structure and comparison of computational tools that predict their utilization. *Nucleic Acids Res.* 34(16):4630-41, 2006.
43. Gruber FX, Hjorth-Hansen H, Mikkola I, Stenke L, Johansen T. A novel Bcr-Abl splice isoform is associated with the L248V mutation in CML patients with acquired resistance to imatinib. *Leukemia.* 20(11):2057-60, 2006.

44. Marco EJ, Bristow J, Cotter PD, Stevenson PD, Pennacchio L, Schwartz CE, Sherr EH. *ARHGEF9*: Identification of a novel X-linked mental retardation and behavior disorder gene. Am. Neurological Association, Poster #S-144, 2006.
45. Sabet A, Li J, Ghandour K, Pu Q, Wu X, Kamholz J, Shy ME, Cambi F. Skin biopsies demonstrate *MPZ* splicing abnormalities in Charcot-Marie-Tooth neuropathy 1B. *Neurology*. 67(7):1141-6, 2006.
46. Oliva CP, L Pisciotto, O Guardamagna, A Bellocchio, P Tarugi, FG Schaap, S Bertolini and S Calandra. A novel sequence variant in *APOA5* gene found in patients with severe hypertriglyceridemia. *Atherosclerosis*, 188(1): 215-217, 2006.
47. Torregrossa R, Gambaro G, Fabris A, Citron L, Mezzabotta F, Lupo A, D'Angelo A, Anglani F. Glial cell-line derived neurotrophic factor: Un gene candidate per la patogenesi del rene con midollare a spugna. (http://www.sigu.net/e107_files/downloads/Comunicazioni/Genetica%20delle%20Malattie%20Complesse.pdf), 2006.
48. Garcia-Blanco, M. Alternative Splicing: Therapeutic Target and Tool. In *Alternative Splicing and Disease*, ed. P. Jeanteur, 44:47-64, Springer, Berlin, 2006.
49. Gaedigk A, JS Leeder. Letter to the Editor. *Clinical Pharmacology & Therapeutics* 80, 558 – 560, 2006.
50. Královičová J, H Lei, I Vořechovský. Phenotypic Consequences of Branchpoint Mutations, *Hum. Mut.* 27: 803 – 813, 2006.
51. Y. Von Kodolitsch, J. Berger, P. K. Rogan. Predicting severity of haemophilia A and B splicing mutations by information analysis *Haemophilia* 12 (3), 258–262, 2006.
52. JS Kern. The molecular basis of Dystrophic Epidermolysis Bullosa: Mutation detection and study of clinical, biochemical, and molecular findings in 29 patients (Dissertation), Albert Ludwigs Universität Freiburg, 2005.
53. Godefroid N, Riveira-Munoz E, Saint-Martin C, Nassogne MC, Dahan K, Devuyst O. A novel splicing mutation in *SLC12A3* associated with Gitelman syndrome and idiopathic intracranial hypertension. *Am J Kidney Dis*. 48(5):e73-79, 2006.
54. Wang J, Sonnerborg A, Rane A, Josephson F, Lundgren S, Stahle L, Ingelman-Sundberg M. Identification of a novel specific *CYP2B6* allele in Africans causing impaired metabolism of the HIV drug efavirenz. *Pharmacogenetics and Genomics* 16(2):191-198, 2006
55. P Smit. Factors determining glucocorticoid sensitivity in man. (Dissertation), Erasmus University, Rotterdam, 2006.
56. Oh KS, Khan SG, Jaspers NG, Raams A, Ueda T, Lehmann A, Friedmann PS, Emmert S, Gratchev A, Lachlan K, Lucassan A, Baker CC, Kraemer KH. Phenotypic heterogeneity in the XPB DNA helicase gene (*ERCC3*): xeroderma pigmentosum without and with Cockayne syndrome. *Hum Mutat*. 27(11):1092-103, 2006.
57. Di Leo E, Magnolo L, Lancellotti S, Croce L, Visintin L, Tiribelli C, Bertolini S, Calandra S, Tarugi P. Abnormal apolipoprotein B pre-mRNA splicing in patients with familial hypobetalipoproteinaemia. *J. Med. Genetics* 44:219-224, 2007.
58. Schonfelder E-M, Knuppel T, Tasic V, Miljkovic P, Konrad M, Wuhl E, Antignac C, Bakkaloglu A, Schaefer F, Weber S, and the ESCAPE Trial Group. Mutations in Uroplakin IIIA are a rare cause of renal hypodysplasia in humans. *Am J. Kidney Dis*. 47: 1004-1012, 2006.
59. E.J. Marco, J. Bristow, P.D. Cotter, R.E. Stevenson, L. Pennacchio, C.E. Schwartz, E.H. Sherr. Identification of a Novel X-Linked Mental Retardation and Behavior Disorder

- Gene. #S-144. American Neurological Association 131st Annual Meeting, October 8-11, 2006 – Chicago, Illinois
60. Douglas DA, Zhong H, Ro JY, Oddoux C, Berger AD, Pincus MR, Satagopan JM, Gerald WL, Scher HI, Lee P, Osman I. Novel mutations of epidermal growth factor receptor in localized prostate cancer. *Front Biosci.* 2006 Sep 1;11:2518-25.
 61. Leverenz JB, Yu CE, Montine TJ, Steinbart E, Bekris LM, Zabetian C, Kwong LK, Lee VM-Y, Schellenberg GD, Bird TD. A novel progranulin mutation associated with variable clinical presentation and tau, *TDP43*, and alpha-synuclein pathology. *Brain*, doi:10.1093/brain/awm069, 2007.
 62. Akiyama M, Titeux M, Sakai K, McMillan JR, Tonasso L, Calvas P, F Jossic, A Hovnanian and H Shimizu. DNA-Based Prenatal Diagnosis of Harlequin Ichthyosis and Characterization of *ABCA12* Mutation Consequences. *Journal of Investigative Dermatology* (2007) **127**, 568–573.
 63. A.M.G. Pasmooij, H.H. Pas, M.C. Bolling, and M.F. Jonkman. Revertant mosaicism in junctional epidermolysis bullosa due to multiple correcting second-site mutations in *LAMB3*. *J Clin Invest.* 2007 May 1; 117(5): 1240–1248.
 64. C. Drögemüller, U. Philipp, B. Haase, A-R Günzel-apel, and T. Leeb. A Noncoding Melanophilin Gene (*MLPH*) SNP at the Splice Donor of Exon 1 Represents a Candidate Causal Mutation for Coat Color Dilution in Dogs. *Journal of Heredity*, doi:10.1093/jhered/esm021, 2007.
 65. Zied Ben Selma, Sebnem Yilmaz, Pierre Olivier Schischmanoff, Astrid Blom, Candan Ozogul, Liliane Laroche and Frédéric Caux. A Novel S115G Mutation of *CGI-58* in a Turkish Patient with Dorfman–Chanarin Syndrome. *Journal of Investigative Dermatology* 2007; doi: 10.1038/sj.jid.5700860
 66. E. Wang, N. Dimova, and F. Cambi. *PLP/DM20* ratio is regulated by hnRNPH and F and a novel G-rich enhancer in oligodendrocytes. *Nucleic Acids Research.* 2007. doi:10.1093/nar/gkm387
 67. J Papp, M. Kovacs, E. Olah. Germline *MLH1* and *MSH2* mutational spectrum including frequent large genomic aberrations in hereditary non-polyposis colorectal cancer families: Implications for genetic testing. *World Journal of Gastroenterology* 2007; *13*(19):2727-2732.
 68. Pasvolsky R, Feigelson SW, Kilic SS, Simon AJ, Tal-Lapidot G, Grabovsky V, Crittenden JR, Amariglio N, Safran M, Graybiel AM, Rchavi G, Ben-Dor S, Etzioni A, Alon R. A LAD-III syndrome is associated with defective expression of the Rap-1 activator CalDAG-GEFI in lymphocytes, neutrophils, and platelets. *J. Exp. Med.* Doi: 10.1084/jem.20070058, 2007.
 69. Keren B, Suzuki OT, Gérard-Blanluet M, Brémond-Gignac D, Elmaleh M, Titomanlio L, Delezoide A-L, Passos-Bueno MR, Verloes A. 2007. CNS malformations in Knobloch syndrome with splice mutation in *COL18A1* gene. *Am J Med Genet Part A* *143A*:1514-1518.
 70. KM Sanggaard, Rendtorff ND, Kjaer KW, Eiberg H, Johnsen T, Gimsing S, Dyrmosé J, Nielsen KO, Lage K and Tranebjærg L. Branchio–oto–renal syndrome: detection of *EYAI* and *SIX1* mutations in five out of six Danish families by combining linkage, MLPA and sequencing analyses. *European Journal of Human Genetics.* 18 July 2007; doi: 10.1038/sj.ejhg.5201900

71. SC Jung, Park JW, Cho DY, Yang JH, Yoon HR, Shetty G, Desai S. PHEX gene mutations and genotype-phenotype analysis of Korean patients with Hypophosphatemic Ricketts. EPOS/IFPOS Combined Meeting, 11-14 Apr 2007, Sorrento, Italy.
72. IM Day, Kralovicova J, Gaunt TR, Rodriguez S, Wood PJ, Vorechovsky I. IDDM2 locus: 5' noncoding intron I splicing and translational efficiency effects of INS - 23HphI – more than a tag for the INS promoter VNTR. *Diabetes* 55[1]:260-264, 2006, and HGM 2006.
73. Simpson MA, Hsu R, Keir LS, Hao J, Sivapalan G, Ernst LM, Zackai EH, Al Gazali LI, Hulskamp G, Kingston HM, Prescott TE, Ion A, Patton MA, Murday V, George A, Crosby AH. Mutations in *FAM20C* are associated with lethal osteosclerotic bone dysplasia (Raine syndrome), highlighting a crucial molecule in bone development *Am J. Hum. Genet.* 81: 906–912, 2007.
74. K. Arita, V. Wessagowit, A.C. Inamadar, A. Palit, H. Fassih, J.E. Lai-Cheong, C. Pourreyaon, A.P. South, J.A. McGrath. Unusual molecular findings in Kindler syndrome. *British Journal of Dermatology*. doi:10.1111/j.1365-2133.2007.08159.x
75. M A. Garcia-Gonzalez, J G. Jones, S K. Allen, C M. Palatucci, S D. Batish, W K. Seltzer, Z Lan, E Allen, F Qian, X M. Lens, Y Pei, GG. Germino and T J. Watnick. Evaluating the clinical utility of a molecular genetic test for polycystic kidney disease. *Molecular Genetics and Metabolism* . 92:160-167, 2007.
76. S-W. OH, J.S. LEE, M.Y. KIM And S-C. KIM. *COL7A1* mutational analysis in Korean patients with dystrophic epidermolysis bullosa. *British Journal of Dermatology*. doi:10.1111/j.1365-2133.2007.08191.x, 2007.
77. E Marco, FE Abidi , J Bristow , WB Dean, PD Cotter , RJ Jeremy , CE. Schwartz and EH Sherr. *ARHGEF9* disruption in a female patient is associated with X linked mental retardation and sensory hyperarousal. *J. Med Genet*. doi:10.1136/jmg.2007.052324, 2007.
78. P. Henneman, F. G. Schaap, P. C. N. Rensen, K. W. van Dijk, A. H. M. Smelt (2008) Estrogen induced hypertriglyceridemia in an apolipoprotein AV deficient patient. *Journal of Internal Medicine* 263 (1), 107–108, 2007.
79. E. Riveira-Munoz, Q. Chang, N. Godefroid, J.G. Hoenderop, R.J. Bindels, K. Dahan, and O. Devuyst and the Belgian Network for the Study of Gitelman Syndrome. Transcriptional and Functional Analysis of *SLC12A3* Mutations: New Clues for the Pathogenesis of Gitelman Syndrome. *J. Am Soc. Nephrol* 18: 1271-83, 2007.
80. Gaedigk A, Gaedigk R, Leeder JS. *CYP2D7* splice variants in human liver and brain: does *CYP2D7* encode functional protein? *Biochem Biophys Res Commun* 336:1241–1250. (2006)
81. Schwaderer P, Knüppel T, Konrad M, Mehls O, Schärer K, Schaefer F, Weber S. Clinical course and *NPHS2* analysis in patients with late steroid-resistant nephrotic syndrome. *Pediatr Nephrol.* 2007 Nov 14, doi: 10.1007/s00467-007-0653-5.
82. S. A. Lietman, J. Goldfarb, N. Desai, M. A. Levine. Preimplantation Genetic Diagnosis for Severe Albright Hereditary Osteodystrophy. *J Clin Endocrin Metab.* Dec. 18, 2007, doi:10.1210/jc.2007-2040.
83. S-W Oh, J S Lee, M Y Kim and S-C Kim . Novel keratin 5 mutations in epidermolysis bullosa simplex: Cases with unusual genotype–phenotype correlation. *J. Derm. Science* 48[3]: 229-232, 2007.

84. K Zhang, I Nowak, D Rushlow, B. L. Gallie, D. R. Lohmann. Patterns of missplicing caused by *RBI* gene mutations in patients with retinoblastoma and association with phenotypic expression. *Hum Mut.* Jan 7, 2008. doi:10.1002/humu.20664.
85. G Caridi, M Dagnino, B Dalgic, O Egritas, B Sancak, M Campagnoli, L Dolcini, M Galliano and L Minchiotti. Analbuminemia Zonguldak: Case report and mutational analysis, *Clinical Biochemistry*, doi:10.1016/j.clinbiochem.2007.11.016, 2007.
86. H-R Song, J-W Park, D-Y Cho, J H Yang, H-R Yoon, S-C Jung. *PHEX* Gene Mutations and Genotype-Phenotype Analysis of Korean Patients with Hypophosphatemic Rickets, *J Korean Med Sci*; 22: 981-6, 2007.
87. J Brockmöller, M V. Tzvetkov. Pharmacogenetics: data, concepts and tools to improve drug discovery and drug treatment. *European Journal of Clinical Pharmacology*, 64[2]: 133-157, 2007.
88. G.H. Kim, J.M. Ko, J.J. Lee, H.W. Yoo. A novel intronic point mutation of *CPS1* gene in a Korean family with CPS1 deficiency. American Society of Human Genetics Meeting, San Diego CA, 2007 (<http://www.ashg.org/genetics/ashg07s/fl0330.htm>)
89. Olga Anczuków, Monique Buisson, Marie-Josèphe Salles, Sarah Triboulet, Michel Longy, Rosette Lidereau, Olga M. Sinilnikova, Sylvie Mazoyer. Unclassified variants identified in *BRCA1* exon 11: Consequences on splicing. *Genes, Chromosomes and Cancer*, 47: 418-426, 2008.
90. Christian Beetz, Rebecca Schüle, Tine Deconinck, Khanh-Nhat Tran-Viet, Hui Zhu, Berry P.H. Kremer, Suzanna G.M. Frints, Wendy A.G. van Zelst-Stams, Paula Byrne, Susanne Otto, Anders O.H. Nygren, Jonathan Baets, Katrien Smets, Bertien Ceulemans, Bernard Dan, Narasimhan Nagan, Jan Kassubek, Sven Klimpe, Thomas Klopstock, Henning Stolze, Hubert J.M. Smeets, Constance T.R.M. Schrandt-Stumpel, Michael Hutchinson, Bart P. van de Warrenburg, Corey Braastad, Thomas Deufel, Margaret Pericak-Vance, Ludger Schöls, Peter de Jonghe and Stephan Züchner. *REEPI* mutation spectrum and genotype/phenotype correlation in hereditary spastic paraplegia type 31, *Brain*, doi:10.1093/brain/awn026, 2008.
91. Sznajer Y, Coldéa C, Meire F, Delpierre I, Sekhara T, Touraine RL. A de novo *SOX10* mutation causing severe type 4 Waardenburg syndrome without Hirschsprung disease. *Am J Med Genet Part A* 146A:1038, 2007.
92. B Borroni, S Archetti, A Albericci, C Agosti, M Gennarelli, B Bigini, C Bonvicini, M Ferrari, G Bellelli, D Galimberti, E Scarpini, D Di Lorenzo, L Caimi, C Caltagirone, M Di Luca. Progranulin genetic variations in frontotemporal lobar degeneration: evidence for low mutation frequency in an Italian clinical series. *Neurogenetics*, 10.1007/s10048-008-0127-3, 2008.
93. Houdayer C, Dehainault C, Mattler C, Michaux D, Caux-Moncoutier V, Pagès-Berhouet S, d'Enghien CD, Laugé A, Castera L, Gauthier-Villars M, Stoppa-Lyonnet D. Evaluation of in silico splice tools for decision-making in molecular diagnosis. *Hum Mutat.* 2008 Apr 30. PMID: 18449911
94. Hiroki Inui, Kyu-Seon Oh, Carine Nadem, Takahiro Ueda, Sikandar G Khan, Ahmet Metin, Engin Gozukara, Steffen Emmert, Hanoch Slor, David B Busch, Carl C Baker, John J DiGiovanna, Deborah Tamura, Cornelia S Seitz, Alexei Gratchev, Wen Hao Wu, Kee Yang Chung, Hye Jin Chung, Esther Azizi, Roger Woodgate, Thomas D Schneider and Kenneth H Kraemer. Xeroderma Pigmentosum-Variant Patients from

- America, Europe, and Asia. *Journal of Investigative Dermatology*, 27 March 2008; doi: 10.1038/jid.2008.48.
95. Hines, RN., Koukouritaki, S B., Poch, M T. and Stephens, M C. (2008) 'Regulatory Polymorphisms and their Contribution to Interindividual Differences in the Expression of Enzymes Influencing Drug and Toxicant Disposition', *Drug Metabolism Reviews*, 40:2, 263 — 301, 2008.
 96. T . Hamada , S . Fukuda , S . Sakaguchi , S . Yasumoto , S . Kim , T . Hashimoto. Molecular and clinical characterization in Japanese and Korean patients with Hailey–Hailey disease: Six new mutations in the *ATP2C1* gene. *Journal of Dermatological Science* , Volume 51 (1) : 31 – 36, 2008.
 97. Riveira-Munoz E, Devuyst O, Belge H, Jeck N, Strompf L, Vargas-Poussou R, Jeunemaître X, Blanchard A, Knoers NV, Konrad M, Dahan K. Evaluating *PVALB* as a candidate gene for *SLC12A3*-negative cases of Gitelman's syndrome. *Nephrol Dial Transplant*. doi:10.1093/ndt/gfn229, 2008.
 98. Chen L, S Qin, J Xie, J Tang, L Yang, W Shen, X Zhao, J Du, G He, G Feng, L He, Q Xing. Genetic polymorphism analysis of *CYP2C19* in Chinese Han populations from different geographic areas of mainland China. *Pharmacogenomics*, 9(6): 691-702, 2008.
 99. Tournier I, M Vezain, A Martins, F Charbonnier , S Baert-Desurmont, S Olschwang, Q Wang , MP Buisine, J Soret, J Tazi , T Frébourg, M Tosi. A large fraction of unclassified variants of the mismatch repair genes *MLH1* and *MSH2* is associated with splicing defects. *Hum Mut*, 2008 DOI: 10.1002/humu.20796.
 100. Bonnet-Dupeyron M-N, P Combes, P Santander, F Cailloux, O Boespflug-Tanguy, C Vauris-Barrière. *PLP1* splicing abnormalities identified in Pelizaeus-Merzbacher disease and SPG2 fibroblasts are associated with different types of mutations. *Human Mutation*, 2008. DOI: 10.1002/humu.20758
 101. Kölsch H, F Jessen, J Wiltfang, P Lewczuk, M Dichgans, J Kornhuber, L Frölich, I Heuser, O Peters, JB Schulz, SG Schwab and W Maier. Influence of *SORL1* gene variants: Association with CSF amyloid- β products in probable Alzheimer's disease. *Neuroscience Letters* 440 (1): 68-71.
 102. Hartmann L, Theiss S, Niederacher D, Schaal H. Diagnostics of pathogenic splicing mutations: does bioinformatics cover all bases? *Front Biosci*. 2008 May 1;13:3252-72.
 103. Luquin N, Yu B , Trent R , Morahan J, Pamphlett R. An analysis of the entire *SOD1* gene in sporadic ALS . *Neuromuscular Disorders*, 18 [7] : 545 - 552, 2008.
 104. Qin S, Shen L, Zhang A, Xie J, Shen W, Chen L, Tang J, Xiong Y, Yang L, Shi Y, Feng G, He L, and Xing Q. Systematic polymorphism analysis of the *CYP2D6* gene in four different geographical Han populations in mainland China. *Genomics*, 2008 doi:10.1016/j.ygeno.2008.05.004
 105. López-Jiménez E, de Campos JM, Kusak EM, Landa I, Leskelä S, Montero-Conde C, Leandro-García LJ, Vallejo LA, Madrigal B, Rodríguez-Antona C, Robledo M, Cascón A. SDHC mutation in an elderly patient without familial antecedents. *Clinical Endocrinology*, 2008, DOI: 10.1111/j.1365-2265.2008.03368.x
 106. Nalla VK, Peter K. Rogan. Erratum: Automated splicing mutation analysis by information theory, *Human Mutation* [Volume 29 Issue 9](#), Page 1168, doi: 10.1002/humu.20867
 107. Alcántara-Ortigoza MA, Belmont-Martínez L, Vela-Amieva M, González-Del Angel A. Analysis of the CTNS Gene in Nephropathic Cystinosis Mexican Patients: Report of

- Four Novel Mutations and Identification of a False Positive 57-kb Deletion Genotype with LDM-2/Exon 4 Multiplex PCR Assay. *Genet Test.* 2008 Sep;12(3):409-14.
108. Vreeswijk MPG, Kraan JN, van der Klift HM, Vink GR, Cornelisse CJ, Wijnen JT, Bakker E, van Asperen CJ, Devilee P. Intronic variants in *BRCA1* and *BRCA2* that affect RNA splicing can be reliably selected by splice-site prediction programs. *Hum. Mut.* Doi:10.1002/humu.20811, 2008.
109. A. Cefalù, D. Noto, L. Magnolo, E. Pinotti, M. Gomaschi, S. Martini, G. Vigna, L. Calabresi, P. Tarugi, M. Averna. Novel mutations of CETP gene in Italian subjects with hyperalphalipoproteinemia. *Atherosclerosis*, doi:10.1016/j.atherosclerosis.2008.08.031, 2008.
110. N. Godefroid, E. Riveira-Munoz, C. Saint-Martin, M. Nassogne, K. Dahan, O. Devuyt. A Novel Splicing Mutation in *SLC12A3* Associated With Gitelman Syndrome and Idiopathic Intracranial Hypertension. *American Journal of Kidney Diseases*, 48[5]: e73 - e79, 2008.
111. Palomino-Doza J., Rahman TJ, Avery PJ, Mayosi BM, Farrall M, Watkins H, Edwards C.R.W., and Keavney B. Ambulatory Blood Pressure Is Associated With Polymorphic Variation in P2X Receptor Genes. *Hypertension.* 52:980-985, 2008.
112. Watnick TJ; Garcia-Gonzalez M; Germino GG; Jones J G. PKD Mutations and evaluation of the same. *Int. Pat. App. No. WO2008094194 (A2).* 2008.
113. Geeta Hampson, Martin A Konrad, and John Scoble. Familial hypomagnesaemia with hypercalciuria and nephrocalcinosis (FHHNC): Compound heterozygous mutation in the claudin 16 (*CLDN16*) gene. *BMC Nephrol.* 9: 12. doi: 10.1186/1471-2369-9-12. 2008.
114. Stefan Bröer, Charles G. Bailey, Sonja Kowalczyk, Cynthia Ng, Jessica M. Vanslambrouck, Helen Rodgers, Christiane Auray-Blais, Juleen A. Cavanaugh, Angelika Bröer and John E.J. Rasko. Iminoglycinuria and hyperglycinuria are discrete human phenotypes resulting from complex mutations in proline and glycine transporters. *J. Clin. Invest.* doi:10.1172/JCI36625, 2008.
115. Shaohua Fang, Xiangming Guo, Xiaoyun Jia, Xueshan Xiao, Shiqiang Li, Qingjiong Zhang. Novel *GPR143* mutations and clinical characteristics in six Chinese families with X-linked ocular albinism, *Molecular Vision* 2008; 14:1974-1982.
116. Barbara Borroni, Silvana Archetti, Antonella Alberici, Chiara Agosti, Massimo Gennarelli, Barbara Bigni, Cristian Bonvicini, Maria Ferrari, Giuseppe Bellelli, Daniela Galimberti, Elio Scarpini, Diego Di Lorenzo, Luigi Caimi, Carlo Caltagirone, Monica Di Luca, Alessandro Padovani. Progranulin genetic variations in frontotemporal lobar degeneration: evidence for low mutation frequency in an Italian clinical series. *Neurogenetics* 9[3], 197-205, 2008. DOI - 10.1007/s10048-008-0127-3
117. Amanda B. Spurdle, Fergus J. Couch, Frans B.L. Hogervorst, Paolo Radice, Olga M. Sinilnikova, for the IARC Unclassified Genetic Variants Working Group. Prediction and assessment of splicing alterations: implications for clinical testing. *Human Mutation.* 29 [11]: 1304 - 1313, 2008.
118. Enza Di Leo, Lucia Magnolo, Elisa Pinotti, Scipione Martini, Irene Cortella, Nicola Vitturi, Claudio Rabacchi, Alessia Wunsch, Francesco Pucci, Stefano Bertolini, Sebastiano Calandra, and Patrizia Tarugi. Functional analysis of two novel splice site mutations of *APOB* gene in familial hypobetalipoproteinemia. *Molecular Genetics and Metabolism*, doi:10.1016/j.ymgme.2008.10.016, 2008.

119. S Bloethner, A Mould, M Stark, NK Hayward. Identification of *ARHGEF17*, *DENND2D*, *FGFR3*, and *RBI* mutations in melanoma by inhibition of nonsense-mediated mRNA decay. *Genes, Chromosomes and Cancer*, 47[12]:1076-1085, 2008.
120. Najah M, E. Di Leo, J Awatef, L Magnolo, J Imene, E Pinotti, M Bahri, S Barsaoui, I Brini, M Fekih, M Slimane and P Tarugi. Identification of patients with abetalipoproteinemia and homozygous familial hypobetalipoproteinemia in Tunisia. *Clinica Chimica Acta*. doi:10.1016/j.cca.2008.11.012, 2008.
121. Elena Botta, Tiziana Nardo, Donata Orioli, Roberta Guglielmino, Roberta Ricotti, Sergio Bondanza, Francesco Benedicenti, Giovanna Zambruno, Miria Stefanini. Genotype-phenotype relationships in trichothiodystrophy patients with novel splicing mutations in the *XPD* Gene. *Human Mutation*, 2008. 10.1002/humu.20912
122. Nikolay Mintchev, Eleni Zamba-Papanicolao, Kleopas A. Kleopa, and Kyproula Christodoulou. A novel *ALS2* splice-site mutation in a Cypriot juvenile-onset primary lateral sclerosis family. *NEUROLOGY* 2009;72:28-32.
123. Cruchaga C, Fernández-Seara MA, Seijo-Martínez M, Samaranch L, Lorenzo E, Hinrichs A, Irigoyen J, Maestro C, Prieto E, Martí-Clement JM, Arbizu J, Pastor MA, Pastor P. Cortical Atrophy and Language Network Reorganization Associated with a Novel Progranulin Mutation. *Cereb Cortex*. 2008 Dec 10. PMID: 19020205
124. American Society of Human Genetics 2008 annual meeting. Abstracts #1352 and 1356.
125. Rhine J, Mantaring MM, Gardner DF, Miller M. Multiple splice defects in *ABCA1* cause low HDL-C in a family with hypoalphalipoproteinemia and premature coronary artery disease. *BMC Medical Genetics* 10:1, 2009.
126. Eckl KM, de Juanes S, Kurtenbach J, Nätebus M, Lugassy J, Oji V, Traupe H, Preil ML, Martínez F, Smolle J, Harel A, Krieg P, Sprecher E, Hennies HC. Molecular Analysis of 250 Patients with Autosomal Recessive Congenital Ichthyosis: Evidence for Mutation Hotspots in *ALOXE3* and Allelic Heterogeneity in *ALOX12B*. *J Invest Dermatol*. 2009 Jan 8. [Epub ahead of print]. PMID: 19131948
127. Liu Z, Venkatesh SS, Maley CC. Sequence space coverage, entropy of genomes and the potential to detect non-human DNA in human samples. *BMC Genomics*. 9:509, 2008.
128. Bogaerts V, K Nuytemans, J Reumers, P Pals, S Engelborghs, B Pickut, E Corsmit, K Peeters, J Schymkowitz, P De Deyn, P Cras, F Rousseau, J Theuns, C Van Broeckhoven. Genetic variability in the mitochondrial serine protease *HTRA2* contributes to risk for Parkinson disease. *Human Mutation* 29:832-40, 2008.
129. Pelucchi S, R Mariani, P Trombini, S Coletti, M Pozzi, V Paolini, D Barisani, A Piperno. Expression of hepcidin and other iron-related genes in type 3 hemochromatosis due to a novel mutation in transferrin receptor-2. *Haematologica*, Vol 94, 276-279 doi:10.3324/haematol.13576.
130. Yu, B. Role of *In Silico* Tools in Gene Discovery. *Molecular Biotechnology*, 41, 296-306, 2009.
131. M. Dua-Awereh, Y. Shimomura, L. Kraemer, M. Wajid, A. Christiano. Mutations in the desmoglein 1 gene in five Pakistani families with striate palmoplantar keratoderma. *Journal of Dermatological Science*, 53[3],192-197, 2009.
132. S Megremis, A Mitsioni, A G. Mitsioni, I Fylaktou, S Kitsiou-Tzelli, C J. Stefanidis, E Kanavakis, J Traeger-Synodinos. Nucleotide Variations in the *NPHS2* Gene in Greek Children with Steroid-Resistant Nephrotic Syndrome (SRNS) Genetic Testing and Molecular Biomarkers., ahead of print. doi:10.1089/gtmb.2008.0083, 2009.

133. H Kölsch; D Lütjohann; F Jessen; J Popp ; F Hentschel ; P Kelemen ; S Friedrichs, W Maier, R Heun. *RXRA* gene variations influence Alzheimer's disease risk and cholesterol metabolism. *Journal of Cellular and Molecular Medicine*. doi: 10.1111/j.1582-4934.2008.00383.x, 2009.
134. L. Wan, C.-C. Lee, C.-M. Hsu, W.-L. Hwu, C.-C. Yang, C.-H. Tsai and F.-J. Tsai: Identification of eight novel mutations of the acid α -glucosidase gene causing the infantile or juvenile form of glycogen storage disease type II. *Journal of Neurology*, 255:831-838, 2008.
135. T Wong, L Gammon, L Liu, J E Mellerio, P J C Dopping-Hepenstal, J Pacy, G Elia, R Jeffery, Irene M Leigh, H Navsaria and J A McGrath: Potential of Fibroblast Cell Therapy for Recessive Dystrophic Epidermolysis Bullosa. *Journal of Investigative Dermatology* (2008) **128**, 2179–2189; doi:10.1038/jid.2008.78
136. A ElSharawy , B Hundrieser , M Brosch , M Wittig , K Huse , M Platzer , A Becker , M Simon , P Rosenstiel , S Schreiber , M Krawczak , J Hampe : Systematic evaluation of the effect of common SNPs on pre-mRNA splicing. *Human Mutation*. doi: 10.1002/humu.20906, 2009.
137. François-Olivier Desmet, Dalil Hamroun, Marine Lalande, Gwenaëlle Collod-Bérout, Mireille Claustres and Christophe Bérout. Human Splicing Finder: an online bioinformatics tool to predict splicing signals. *Nucleic Acids Research*, doi:10.1093/nar/gkp215, 2009.
138. Heike Kölsch, Dieter Lütjohann, Frank Jessen, Julius Popp, Frank Hentschel, Peter Kelemen, Sandra Schmitz, Wolfgang Maier and Reinhard Heun. *CYP46A1* variants influence Alzheimer's disease risk and brain cholesterol metabolism. *European Psychiatry*, 24(3): 183-190, 2009.