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HUMAN GENE MUTATIONS

Novel human pathological mutations

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Gene symbol: HEXA

Disease: Tay-Sachs disease

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Input for Missense/Nonsense Mutations (*single base-pair substitutions*)

Accession	Codon number	Nucleotide substitution	Amino acid substitution
HM080090	497	TAT-TGT	Tyr-Cys

Gene symbol: SLC3A1

Disease: Cystinuria

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Input for Missense/Nonsense Mutations (*single base-pair substitutions*)

Accession	Codon number	Nucleotide substitution	Amino acid substitution
HM080091	397	TAT-TGT	Tyr-Cys

Gene symbol: SLC3A1

Disease: Cystinuria

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Input for Missense/Nonsense Mutations (single base-pair substitutions)

Accession	Codon number	Nucleotide substitution	Amino acid substitution
HM080092	584	aAGA-TGA	Arg-Term

Gene symbol: SLC7A9

Disease: Cystinuria

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Input for small deletions (<21 bp)

Accession	Deletion	Codon number/location
HD080027	CCAAGGA^AACacAAAGAATTTT	203

Gene symbol: ABCA4

Disease: Macular dystrophy

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Input for Missense/Nonsense Mutations (single base-pair substitutions)

Accession	Codon number	Nucleotide substitution	Amino acid substitution
HM080093	187	CGT-CAT	Arg-His

Gene symbol: UBE3A

Disease: Angelman Syndrome

Evmorfia Tzagkaraki, Sofocleous Christalena, Fryssira Helen, Dinopoulos Argyris, Mavrou Ariadni, Kanavakis Emmanuel
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Input for small insertions (*<21 bp*)

Accession	Insertion	Codon number/location
HI080014	GCTGAG^GCATgTGGTACAGAG	139

Comments: The mutation was detected by ECMA (Enzymatic Cleavage Mismatch Analysis) and characterized by direct sequencing (performed twice).The proband is a 27 months boy with microcephaly and presents a typical for Angelman EEG. Mutation analysis for both parents revealed normal sequences. Sequencing results available upon request.

Gene symbol: JAG1

Disease: Allagille syndrome

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Input for small insertions (*<21 bp*)

Accession	Insertion	Codon number/location
HI080015	TCCTCCAG_I16E17_GTt^GACAGTCAGT	706; c.2115dupT; p.Asp706Stop

Comments: cacgt(T)gaca T is the inserted base.

Gene symbol: COL4A5

Disease: Alport syndrome

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Input for small deletions (*<21 bp*)

Accession	Deletion	Codon number/location
HD080028	CTTACTG^GCCctGAGTCTTTGG	17

Comments: Female with asymptomatic hematuria and proteinuria. c.49_50delCT/p.Leu17GlufsX22.

Gene symbol: F9

Disease: Haemophilia B

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Input for small insertions (*<21 bp*)

Accession	Insertion	Codon number/location
HI080016	GTGGTT^TGCTcctgctCCTGTACTGA	109

Comments: Novel insertion in Pakistani patient.

Gene symbol: F8

Disease: Haemophilia A

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Input for small deletions (*<21 bp*)

Accession	Deletion	Codon number/location
HD080029	GCTCAA^ACACtCTTGATGGAC	298

Comments: Novel deletion in a Pakistani patient.

Gene symbol: RHD

Disease: Rhesus negative blood group

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Input for complex rearrangements

Accession	Description
HP080001	Hybrid with ex. 4-9 RHCE

Comments: Haplotype—cdE.

Gene symbol: ALAS2

Disease: Sideroblastic anaemia

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Input for Missense/Nonsense Mutations (*single base-pair substitutions*)

Accession	Codon number	Nucleotide substitution	Amino acid substitution
HM080094	503	GCC-GTC	Ala-Val

Comments: Mutation found in the propositus and his mother.

Gene symbol: SRY

Disease: XY sex reversal

Celia Ravel, B. Lakhal, H. Elghezal, R. Braham, A. Saad, A. Bashamboo, J.P. Siffroi, K. McElreavey, S. Christin-Maitre
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Input for regulatory mutations

Accession	Nucleotide substitution	Location relative to
HR080003	TGGTTGGGCGGGGTTGAGGGGGTGTTGAGG(G-C) CGGAGAAATGCAAGTTTCATTACAAAAGTT	Initiator methionine

Comments: A 34 year-old phenotypically normal female was referred to our attention for primary amenorrhea. The vagina was present but reduced in length. The weight was 69 kg; the height 1 m 69. FSH, LH, estradiol, prolactin and testosterone levels reached 110 mUI/ml, 24.3 mUI/ml, 12 pg/ml, 12.6 ng/ml and 0.3 ng/ml, respectively. Karyotype was 46,XY. At ultrasound examination, gonads could not be identified. However, a small uterus was present. The two kidneys were normal. SRY mutational analysis revealed a single base-pair substitution. c. -130G > C located in a highly conserved Sp1A motif that has previously been shown experimentally to be involved in regulation of SRY expression. tgttgagg(g-c)cggagaaa.

Gene symbol: APC

Disease: Adenomatous polyposis coli

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Input for splicing mutations (*single base-pair substitution*)

Accession	Intron designation, number or letter	Donor/Acceptor	Relative location	Nucleotide substitution
HS080016	14	Donor	+1	G–C

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