Correction to Hm0516b published in Hum Genet (2005) Vol. 118, page 536

**Gene Symbol:** SCN5A

Disease: Brugada syndrome

E. Arbustini, M.F. Scaffino, M. Diegoli, N. Marziliano, M. Grasso, M. Pasotti, P. Baraldi, R.G. Zennaro

It should read N. Marziliano instead of N. Maziliano

**Gene Symbol:** ALMS1

Disease: Alstrom syndrome

K.J. Flintoff

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Odile Boute-Benejean

Service de Genetique Clinique, Hopital Jeanne Flandre, 59037 Lille Cedex, France

**Small Deletions (<21 bp)**

| Accession number | Codon number/location | Deletion |
|------------------|-----------------------|----------|
| Hd0701           | 2098                  | ATTTT^CAcaGAGAG |

**Comments:** c.6294_6295delCA

**Gene Symbol:** APOB

Disease: Normotriglyceridemic hypobetalipoproteinemia

Vivienne Homer, Peter M. George, Stephen du Toit, James S. Davidson, Callum J. Wilson

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**Small Deletions (<21 bp)**
Accession number:  
Hd0702

Codon number/location:  
3023

Deletion:  
GCATCC^acaaACAATGAA

Comments: Reference:
Mental retardation and ataxia due to normotriglyceridemic hypobetalipoproteinemia. 
Ann Neurol. 2005 Jul;58(1):160–163. 
PMID: 15984016 [PubMed—indexed for MEDLINE]

Gene Symbol: G6PD

Disease: Glucose-6-phosphate dehydrogenase deficiency

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Small Deletions (<21 bp)

Accession number:  
Hd0703

Codon number/location:  
359

Deletion:  
TGC^GgcaaggccctgaacgagcGCAAG

Comments: The short deletion of 18 nucleotides corresponds to [del c.1076–1094] within exon 10 resulting in the 6 amino acids deletion [del p. 359–364] near the dimer interface of the G6PD enzyme. The new mutation was named G6PD Tondela after the patient place of birth.

Gene Symbol: FECH

Disease: Porphyria, erythropoietic

V. Brancalonei, E. Di Pierro, V. Besana, S. Ausenda, S. Drury, M.D. Cappellini
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Gross Deletions

Accession number:  
Hg0701

Deletion: The first breakpoint was located in the intron 1 of the FECH gene at nucleotide 53402361, the second was located in the intragenic space at nucleotide 53412738. The 53402361-53412738del10377 bp causes the loss of the promoter and of the exon 1 of the FECH gene.

Description: DNA analysis revealed the presence of a 10,377 bp

Comments: GeneBank Accession Number NC_000018, CON 30-AUG-2006

Version: NC_000018.8 GI:51511735

Gene Symbol: TAP2

Disease: HLA class I deficiency

Henri de la Salle, Dominique Fricker, Daniel Hanau, Figen Dogu, Aydan Ikinciogullari
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Springer
Small Insertions (<21 bp)

| Accession number: | Codon number/location: | Insertion: |
|-------------------|-------------------------|------------|
| Hi0701            | 341                     | GCTGCAG^ACCcGTTCGCAGTTT |

**Comments:** Associated to HLA haplotype: A*26 B*08 C*w7 DRB1*15 DQB1*05

**Gene Symbol:** RS1

Disease: X-linked juvenile retinoschisis

**Rosa Riveiro-Alvarez, M.J. Trujillo-Tiebas, A. Gimenez, D. Cantalapiedra, E. Vallespin, J. Aguirre-Lamban, C. Villaverde, C. Ayuso**

Fundacion Jimenez Diaz, Department Genetics, Reyes Catolicos 2, 28040 Madrid, Spain, E-mail: rriveiro@fjd.es, Tel.: +34-915504872

**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0701            | 192           | CCC–CTC                  | Pro–Leu                  |

**Gene Symbol:** ALMS1

Disease: Alstrom syndrome

**Kimberley Flintoff**

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**Richard Paisley**

Consultant Physician, Torbay District General Hospital, Newton Road, TQ2 7AA, UK

**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0702            | 3001          | tCAA–TAA                 | Gln–Term                 |

**Comments:** c.9001C > T, p.Q3001X

**Gene Symbol:** NF1

Disease: Neurofibromatosis 1

**Sing-Chung Li, Cheng-Hung Huang, Kun-Long Hung**

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**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0703            | 553           | tCAT–CGT                 | His–Arg                  |
Gene Symbol: ABCA4

Disease: Stargardt disease

Jana Aguirre-Lamban, R. Riveiro-Alvarez, D. Cantalapiedra, E. Vallespin, M.J. Trujillo-Tiebas, C. Villaverde, C. Ayuso
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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|-------------------------|
| Hm0705           | 234          | cCAG–TAG                | Gln–Stop                |

Gene Symbol: G6PD

Disease: Glucose-6-phosphate dehydrogenase deficiency

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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|-------------------------|
| Hm0706           | 198          | CGC–CAC                 | Arg–His                 |

Gene Symbol: CHM

Disease: Choroideraemia

Cristina Villaverde, M.J. Trujillo-Tiebas, M. Garcia-Hoyos, N. Perez, R.C. Narvaiza, E. Guillén, C. Ayuso
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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|-------------------------|
| Hm0707           | 76           | gCAA–TAA                | Gln–Stop                |

Gene Symbol: CHM

Disease: Choroideraemia

Cristina Villaverde, M.J. Trujillo-Tiebas, M. Garcia-Hoyos, R.C. Narvaiza, N. Perez, B. Garcia-Sandoval, C. Ayuso
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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|-------------------------|
| Hm0708           | 103          | TATg–TAG                | Tyr–Stop                |
Gene Symbol: NOTCH3

Disease: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL)

Susana Ferreira
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Cristina Costa
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João Paulo Oliveira
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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|------------------------|
| Hm0709           | 1099         | TGC–TAC                 | Cys–Tyr                |

Comments: Mutation identified in a 58-year-old female patient, with recurrent ischaemic strokes, in subcortical topography. Autosomal dominant family history of recurrent strokes. Cerebral MRI suggestive of CADASIL. Muscular biopsy showing electron-dense vascular deposits.

Gene Symbol: NOTCH3

Disease: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL)

Rosa S. Silva
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João Paulo Oliveira
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Missense/nonsense mutations (single base-pair substitutions)

| Accession number | Codon number | Nucleotide substitution | Amino acid substitution |
|------------------|--------------|-------------------------|------------------------|
| Hm0710           | 568          | TGT–TAT                 | Cys–Tyr                |

Gene Symbol: NOTCH3

Disease: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL)

Paulo Fontoura, Rui Guerreiro
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João Paulo Oliveira
Consulta de Genética Médica, Hospital São João, Porto, Portugal
Missense/nonsense mutations (single base-pair substitutions)

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0711            | 978           | cAGC–CGC                 | Ser–Arg                  |

**Comments**: Mutation identified in a 64-year-old female patient with cerebrovascular disease, dementia, epilepsy and psychiatric problems. Had first stroke in the fifth decade of life. Cerebral MRI with white matter abnormalities.

**Gene Symbol**: FZD4

Disease: Familial exudative vitreoretinopathy

**Véronique Vieira, Guillaume de la Houssaye, Anouk Dansault, Elodie Perez, Olivier Roche, Jean-Louis Dufler, Cécile Marsac, Maurice Menasche, Marc Abitbol**

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Missense/nonsense mutations (single base-pair substitutions)

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0712            | 499           | cAAA–GAA                 | Lys–Glu                  |

**Comments**: This mutation was found in three members of a family. The two children presented typical clinical features of FEVR whereas the mother is asymptomatic. This is a FZD4 mutation affecting the Lys–Thr–X–X–X–Trp motif which is involved in the activation of Wnt/beta catenin signalling.

**Gene Symbol**: TPI1

Disease: Triosephosphate isomerase deficiency

**L. Manco, M.L. Ribeiro**

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Missense/nonsense mutations (single base-pair substitutions)

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0713            | 62            | GCT–GAT                  | Ala–Asp                  |

**Comments**: A previously undescribed mutation in exon 2 of the TPI gene, the transversion 188 C > A predicting the amino acid change 62 Ala > Asp, was identified in a patient with Triosephosphate Isomerase deficiency, compound heterozygous with the second mutation 104 Glu > Asp. The drastic non-conservative replacement of the nonpolar Ala by the polar acidic residue Asp, and the evolutionary conservation of Ala 62 from C. elegans to humans indicates that this mutation certainly affects the TPI enzymatic activity.

**Gene Symbol**: GUCY2D

Disease: Early onset retinitis pigmentosa

**A. Avila-Fernandez, E. Vallespin, D. Cantalapiedra, R. Riveiro-Alvarez, A. Gimenez, M.J. Trujillo-Tiebas, C. Ayuso**

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**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0714            | 587           | CTC→CGC                  | Leu→Arg                  |

**Gene Symbol:** EPM2A

**Disease:** Lafora progressive myoclonus epilepsy

M.J. Trujillo-Tiebas, M. Fenollar-Cortés, P. Gómez-Garré, I. Lorda-Sánchez, J.M. Serratosa, C. Ayuso García  
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**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0716            | 278           | TGCg→TGA                 | Cys→STOP                 |

**Comments:** The mutation is located in nt.834 in exon 4. The other mutation was W165X in codon 165 (Ianzano et al. 2004). The patient is Spanish.

**Gene Symbol:** TAP2

**Disease:** HLA class I deficiency

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**Missense/nonsense mutations (single base-pair substitutions)**

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|---------------|--------------------------|--------------------------|
| Hm0717            | 623           | aCGA→TGA                 | Arg→Stop                 |

**Comments:** Associated to HLA haplotype:  
A*01 B*08 C*07 DRB1*0701 DQB1*02

**Gene Symbol:** NOTCH3

**Disease:** Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy

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Filipa Malheiro  
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Joaão Paulo Oliveira  
Hospital São João, Porto, Portugal
Missense/nonsense mutations (single base-pair substitutions)

| Accession number: | Codon number: | Nucleotide substitution: | Amino acid substitution: |
|-------------------|--------------|--------------------------|--------------------------|
| Hm0718            | 577          | cACA–GCA                 | Thr–Ala                  |

**Comments:** Mutation not found in a sample of 100 Portuguese healthy individuals.