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CLINICAL UTILITY GENE CARD

Clinical utility gene card for: *MAN1B1* defective congenital disorder of glycosylation

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1. Name of the Disease (Synonyms):
Deficiency of Golgi mannosyl-oligosaccharide α1,2-mannosidase, MAN1B1 deficiency, MAN1B1-CDG.

2. OMIM# of the Disease:
614202.

3. Name of the Analysed Genes or DNA/Chromosome Segments:
MAN1B1.

4. OMIM# of the Gene(s):
604346.

Review of the analytical and clinical validity as well as of the clinical utility of DNA-based testing for mutations in *MAN1B1* in diagnostic, predictive and prenatal settings, and for risk assessment in relatives.

This is the abstract of a paper that is published in full online at www.nature.com/ejhg and can be cited with *European Journal of Human Genetics* (2016) **24,** doi:10.1038/ejhg.2015.248

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CLINICAL UTILITY GENE CARD UPDATE

Clinical utility gene card for: Biotinidase deficiency—update 2015

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1. Name of the Disease (Synonyms):
Biotinidase deficiency (late-onset multiple carboxylase deficiency; late-onset biotin-responsive multiple carboxylase deficiency; juvenile-onset multiple carboxylase deficiency; BTD deficiency)

2. OMIM# of the Disease:
253260

3. Name of the Analysed Genes or DNA/Chromosome Segments:
BTD

4. OMIM# of the Gene(s):
609019

Review of the analytical and clinical validity as well as of the clinical utility of DNA-based testing for variants in the *BTD* gene in diagnostic, predictive and prenatal settings and for risk assessment in relatives.

This is the abstract of a paper that is published in full online at www.nature.com/ejhg and can be cited with *European Journal of Human Genetics* (2016) **24,** doi:10.1038/ejhg.2015.246

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CORRIGENDA

Lessons from a pair of siblings with BPAN

Yuri A Zarate, Julie R Jones, Melanie A Jones, Francisca Millan, Jane Juusola, Annette Vertino-Bell, G Bradley Schaefer and Michael C Kruer

European Journal of Human Genetics (2016) **24,** 1095; doi:10.1038/ejhg.2015.274

Correction to: *European Journal of Human Genetics* (2016) **24**, 1080–1083; doi:10.1038/ejhg.2015.242; published online 18 November 2015

Since the publication of the above paper the authors have realised that they had omitted an important reference published online in this journal while their own paper was undergoing the review process.
In the paper by Abidi *et al.*, the authors describe another case of a male patient with a non-mosaic deletion that encompassed the *WDR45* gene. This reference supports the notion, also demonstrated in their paper, of the viability of non-mosaic males affected with either intragenic mutations or whole-gene deletions affecting the *WDR45* gene. The reference is listed below:
Abidi A, Mignon-Ravix C, Cacciagli P, Girard N, Milh M, Villard L: Early-onset epileptic encephalopathy as the initial clinical presentation of *WDR45* deletion in a male patient. *Eur J Hum Genet*; e-pub ahead of print 15 July 2015; doi:10.1038/ejhg.2015.159.

Genome-wide association studies identify genetic loci for low von Willebrand factor levels

Janine van Loon, Abbas Dehghan, Tang Weihong, Stella Trompet, Wendy L McArdle, Folkert W Asselbergs, Ming-Huei Chen, Lorna M Lopez, Jennifer E Huffman, Frank WG Leebeek, Saonli Basu, David J Stott, Ann Rumley, Ron T Gansevoort, Gail Davies, James JF Wilson, Jacqueline CM Witteman, Xiting Cao, Anton JM de Craen, Stephan JL Bakker, Bruce M Psaty, John M Starr, Albert Hofman, J Wouter Jukema, Ian J Deary, Caroline Hayward, Pim van der Harst, Gordon DO Lowe, Aaron R Folsom, David P Strachan, Nicolas Smith, Moniek PM de Maat and Christopher O'Donnell

European Journal of Human Genetics (2016) **24**, 1096; doi:10.1038/ejhg.2016.21

Correction to: *European Journal of Human Genetics* (2016) **24**, 1035–1040; doi:10.1038/ejhg.2015.222; published online 21 October 2015

Post online publication it was realised that one of the author names (Folkert W Asselbergs) had been submitted incorrectly. This has now been rectified. The corrected paper also appears in this issue.

International Charter of principles for sharing bio-specimens and data

Deborah Mascalzoni, Edward S Dove, Yaffa Rubinstein, Hugh JS Dawkins, Anna Kole, Pauline McCormack, Simon Woods, Olaf Riess, Franz Schaefer, Hanns Lochmüller, Bartha M Knoppers and Mats Hansson

European Journal of Human Genetics (2016) **24**, 1096; doi:10.1038/ejhg.2015.237

Correction to: *European Journal of Human Genetics* (2015) **23**, 721–728; doi:10.1038/ejhg.2014.197; published online 24 September 2014

The authors have requested that Hanns Lochmüller's affiliation be amended to: The John Walton Muscular Dystrophy Research Centre, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, Newcastle University, Newcastle upon Tyne, UK.

Big Data in medical research and EU data protection law: challenges to the consent or anonymise approach

Menno Mostert, Annelien L Bredenoord, Monique CIH Biesaart and Johannes JM van Delden

European Journal of Human Genetics (2016) **24**, 1096; doi:10.1038/ejhg.2016.71

Correction to: *European Journal of Human Genetics* (2016) **24**, 956–960; doi:10.1038/ejhg.2015.239; published online 11 November 2015

This paper has been changed from 'Review' to 'Policy' since advance online publication.