



Corrigendum

Corrigendum to “Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases” [EBioMedicine 54 (2020) 102730]



Jing Tan^{a,b,1,2}, Matias Wagner^{a,c,d,1,*}, Sarah L. Stenton^{a,c}, Tim M. Strom^{a,c},
Saskia B. Wortmann^{a,c,e}, Holger Prokisch^{a,c}, Thomas Meitinger^{a,c}, Konrad Oexle^h,
Thomas Klopstock^{b,f,g,**}

^a Institute of Human Genetics, School of Medicine, Technische Universität München, Munich, Germany

^b Friedrich-Baur-Institute, Department of Neurology, University Hospital, LMU Munich, Munich, Germany

^c Institute of Human Genetics, Helmholtz Zentrum München, Neuherberg, Germany

^d Institute of Neurogenomics, Helmholtz Zentrum München, Neuherberg, Germany

^e Department of Pediatrics, University Children's Hospital, Paracelsus Medical University (PMU), Salzburg, Austria

^f German Center for Neurodegenerative Diseases (DZNE), Munich, Germany

^g Munich Cluster for Systems Neurology (SyNergy), Munich, Germany

^h Institute of Neurogenomics, Neurogenetic Systems Analysis Unit, Helmholtz Zentrum München, Neuherberg, Germany

The authors regret that the published manuscript contains a few rounding and copying errors. The mistakes are minor but affect the allele frequencies of disease-causing variants in 3 genes. These minor corrections do not change the overall description, interpretation, or conclusions of the manuscript.

page 4, paragraph 3.1., 2nd sentence:

The sum of their allele frequencies was 0.0111 in the European (Non-Finnish) and 0.0091 in the total gnomAD dataset resulting in a calculated PKU lifetime risk of 16.0 (14.5–17.6 95% CI)/100,000 in the European (Non-Finnish) population and of 8.2 (7.6–8.9)/100,000 in the total dataset.

is corrected to

The sum of their allele frequencies was 0.0126 in the European (Non-Finnish) and 0.0091 in the total gnomAD dataset resulting in a calculated PKU lifetime risk of 16.0 (14.5–17.6 95% CI)/100,000 in the European (Non-Finnish) population and of 8.2 (7.6–8.9)/100,000 in the total dataset.

page 4, paragraph 3.2., close to the end of 3rd paragraph

The allele frequency of *POLG* mutations was 0.0083 in the European (Non-Finnish) gnomAD dataset, 0.0061 in the total gnomAD

dataset and 0.0074 in the in-house database, resulting in lifetime risk estimates for *POLG*-associated disorders of 6.9 (6.1–7.8), 3.7 (3.4–4.0) and 3.7 (3.3–4.1)/100,000, respectively.

is corrected to

The allele frequency of *POLG* mutations was 0.0083 in the European (Non-Finnish) gnomAD dataset, 0.0061 in the total gnomAD dataset and 0.0061 in the in-house database, resulting in lifetime risk estimates for *POLG*-associated disorders of 6.9 (6.1–7.8), 3.7 (3.4–4.0) and 3.7 (3.3–4.1)/100,000, respectively.

page 6, right column, line 4–6

Implying a combined frequency of 0.0083, and leading to a calculated MCADD lifetime risk of 4.8 (3.6–6.3)/100,000.

is corrected to

Implying a combined frequency of 0.0069, and leading to a calculated MCADD lifetime risk of 4.8 (3.6–6.3)/100,000.

In addition, the Author Contribution section was accidentally omitted due to a production error. This reads:

Author contributions

MW and TK conceived the study. JT and MW defined a comprehensive list of mitochondrial disease genes and set up a list of pathogenic variants in these genes, supported by SLS, TMS, and SBW. JT and MW queried two databases (gnomAD and in house) to assess the allele frequencies of disease-causing variants in the general population and calculated the lifetime risks, supported by HP, TM, KO and TK. JT and MW drafted the manuscript which was then refined by all other authors and finalized by MW and TK.

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* Corresponding author at: Institute of Human Genetics, School of Medicine, Technische Universität München, Munich, Germany.

** Corresponding author at: Friedrich-Baur-Institute, Department of Neurology, University Hospital, LMU Munich, Munich, Germany.

E-mail addresses: matias.wagner@mri.tum.de (M. Wagner), tklopsto@med.lmu.de (T. Klopstock).

¹ These authors contributed equally.

² Present address: Department of Neurology, the Second Affiliated Hospital of Dalian, Medical University, Dalian, China

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