

Defective minor spliceosome mRNA processing results in isolated familial growth hormone deficiency

Jesús Argente, Raquel Flores, Armand Gutiérrez-Arumí, Bhupendra Verma, Gabriel Á Martos-Moreno, Ivon Cuscó, Ali Oghabian, Julie A Chowen, Mikko J Frilander & Luis A Pérez-Jurado

Correction to: *EMBO Mol Med* (2014) 6: 299–306. DOI 10.1002/emmm.201303573 | Published online 30 January 2014

The authors recently noticed that one of the reported mutations, chr1:104093621C>A/p.P474T was incorrectly identified at the cDNA level as c.1320C>A in the text and as c.1302C>A in Fig 2B. The nomenclature at the protein level was correct (p.P474T), and the error should not affect the results or conclusions of the manuscript. In all occurrences throughout the article (including in figure 2), the correct nomenclature of the mutation/gene variant should be c.1420C>A [full nomenclature: NM_017619.4(RNPC3):c.1420C>A (p.Pro474Thr/ p.P474T)].

This genetic variant is correctly listed in ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/variation/587367/>) has an allele frequency

of 0.00003185 in gnomAD (https://gnomad.broadinstitute.org/variant/1-104093621-C-A?dataset=gnomad_r2_1) and has been reported in a second family with growth hormone deficiency due RNPC3 dysfunction (Verberne *et al*, 2020).

Reference

Verberne EA, Faries S, Mannens MMAM, Postma AV, van Haelst MM (2020) Expanding the phenotype of biallelic RNPC3 variants associated with growth hormone deficiency. *Am J Med Genet A* <https://doi.org/10.1002/ajmg.a.61632>

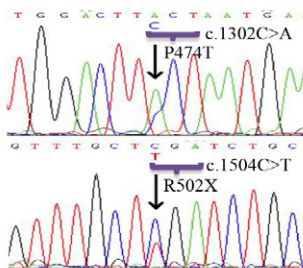


Figure 2B. Original figure.

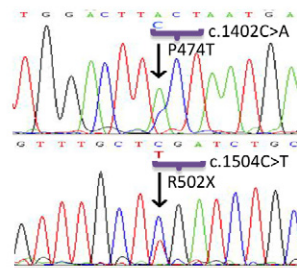


Figure 2B. Corrected figure.