


Response to growth hormone in patients with *RNPC3* mutations

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Correction to: *EMBO Mol Med* (2018) 10: e9143. DOI 10.15252/emmm.201809143 | Published online 6 June 2018

The authors recently noticed that one of the reported mutations, chr1:104093621C>A/p.P474T, was incorrectly listed as c.1320C>A at the cDNA level. The correct mutation is c.1420C>A; the nomenclature at the protein level was correct.

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>