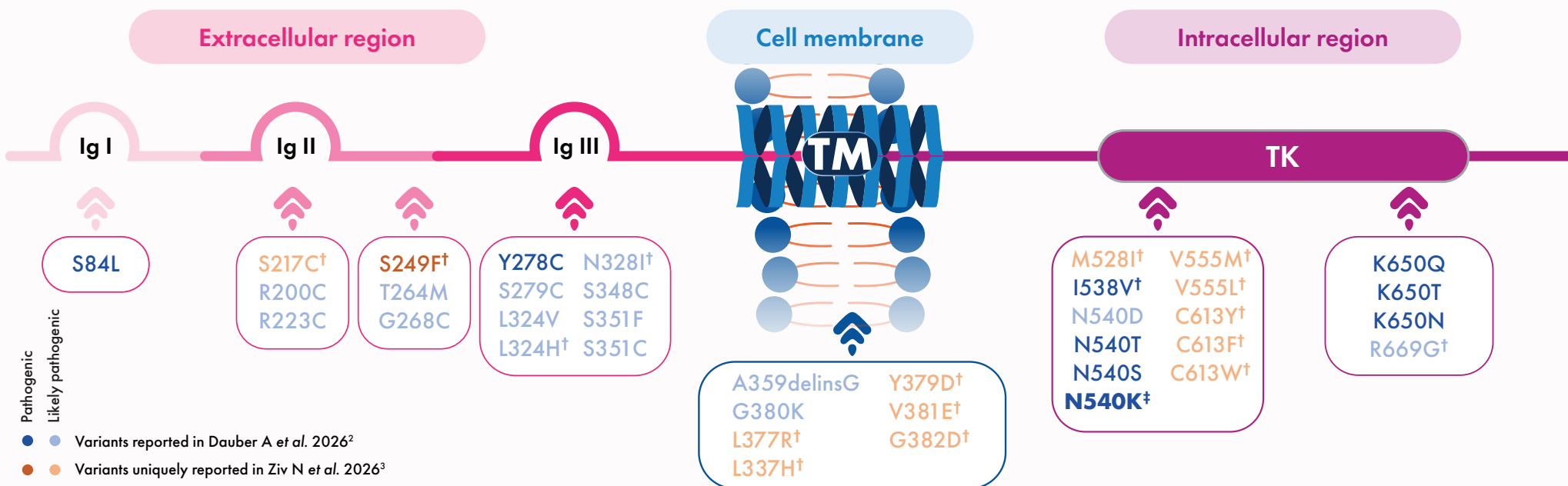
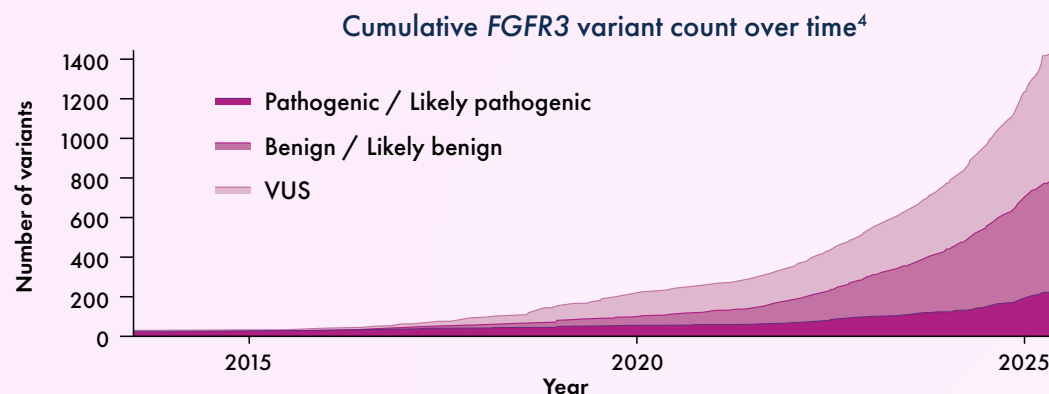


Hypochondroplasia is caused by gain-of-function variants in *FGFR3*^{1-4*}



There are currently 222 pathogenic or likely pathogenic variants identified in *FGFR3*⁴



If you have a scientific or medical information request regarding an *FGFR3* variant not shown here, please contact BioMarin Medical Information: medinfo@bmrn.com. Please do not provide any patient-identifiable information.

*Variants represent the pathogenic / likely pathogenic variants in *FGFR3* associated with hypochondroplasia. These variants do not definitively differentiate between achondroplasia and hypochondroplasia; differentiation is determined by a combination of clinical, radiographic, and genetic observations. [†]These variants are identified as 'novel'. [‡]N540K is the most common *FGFR3* genetic variant in hypochondroplasia, occurring in ~60% of cases.⁵

FGFR3, fibroblast growth factor receptor 3; **Ig**, immunoglobulin; **TK**, tyrosine kinase domain; **TM**, transmembrane domain; **VUS**, variant of uncertain significance.

1. Bober MB et al. Hypochondroplasia. In: GeneReviews® [Internet]. 1999 (updated September 2025). Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1477/>. Accessed August 2026.

2. Dauber A et al. Nat Rev Endocrinol 2026; Epub ahead of print (DOI: 10.1038/s41574-026-01296-z). 3. Ziv N et al. Poster 107 presented at ACMG 2026; Baltimore, MD, USA, 10–14 March 2026.

4. ClinVar. *FGFR3*. Available at: <https://www.ncbi.nlm.nih.gov/clinvar/?gene=FGFR3&term=%22FGFR3%22%5BGENE%5D>. Accessed August 2026. 5. Ornitz DM and Legeai-Mallet L. Dev Dyn 2017; 246 (4): 291–309.

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