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Consulting for a combination of molecular defects for variable expression

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Background: Expressivity is variable for most of the molecular defects. However, achondroplasia is a well-defined and common bone dysplasia, with an incidence of approximately 5-15 per 100,000 live births. Gain-of function mutations in FGFR3 have been shown to cause both chondrodysplasias and craniosynostoses and to result in impaired endochondral ossification.

Case Presentation: A 2-year-old boy with clinical features consistent with achondroplasia and Silver-Russell syndrome-like symptoms was studied. The patient exhibited features such as scoliosis and a trident configuration of the hands, all of which can be explained by a mutations in FGFR3 at c.1138 G >A (p.Gly380Arg). However, prenatal onset growth delay, the speech delay, hypotonia and small triangular face phenotypes were not commonly reported in previous cases of ACH. We further detected a three-fold increase in GRB10 expression. Combining with previous other studies, the one unique feature of this patient that can be directly linked to a GRB10 duplication is the prenatal onset growth delay.

Discussion: The data related to the patient described in the present study at least suggest that mutations in FGFR3 cause ACH, but do not influence the effects of the duplication of GRB10 on prenatal onset growth delay in SRS. The results of our study also suggest that phenotypes are rarely "simple" or directly related to specific gene defects and that combinations of uncommon, rare and exceptional molecular defects, which can be explored and used in diagnoses, may explain the so-called variability observed in the expression of dominant traits.

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