Commentary

Genetic and Molecular Characterization with Robinow Syndrome

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Robinow syndrome (RS) is a rare genetic disorder characterized by limb shortening, craniofacial/orodental abnormalities, and genital hypoplasia. Genetic studies have linked the autosomal recessive form of the disorder to the ROR2 gene on position 9 of the long arm of chromosome 9. The gene is responsible for aspects of bone and cartilage growth. This same gene is involved in causing autosomal dominant brachydactyly B.

Diagnosis for Robinow syndrome is suspected by family history and clinical findings confirmed by typical ROR-2 biallelic pathogenic variants identified by molecular genetic testing. Treatment for various manifestations will usually be addressed by a multidisciplinary team.