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Trio-based Whole Genome Sequencing Arrests de novo Mutations in Pai Syndrome

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ABSTRACT Pai syndrome (PS) is a multiple phenotypic rare syndrome of cleft lip and palate with an unclear diagnostic criteria and etiology. This research reported the clinical phenotypes for two cases of PS and collected the two trios families. Genomic DNA was extracted from blood and sequenced by whole-genome sequencing (WGS) on two probands and their parents. Variant Effect Predictor (VEP) was utilized to determine causative variants of the patients. Identified causal mutations were further confirmed by Sanger Sequencing. WGS analysis identified 153 single-nucleotide polymorphisms (SNPs) and 69 indels in patient 1, while 165 SNPs and 57 indels were identified in patient 2. After multiple filtrations, the researchers identified four de novo missense mutations (FLG, DHX57, ANXA7, and GOLGA3) in patient 1 and two de novo missense mutations (PRSS23 and USP7) in patient 2. These findings still need to be validated but already provide more information on the genetic basis.