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Chromosome and p63 Gene Analysis of an Infant with Ectrodactyly – Split Hand and Foot Malformation

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Abstract: Aim: Split hand-split foot malformation (SHFM) results from central ray reduction and presents as median clefts of the hands and feet, syndactyly, aplasia or hypoplasia of the phalanges, metacarpals and metatarsals, which are frequently associated with other phenotypic abnormalities. We aimed to investigate the genetic pathway of SHFM in a child. Materials and methods: Cytogenetic and molecular genetic analysis was performed in a 10-day-old boy with split-syndactyly hand and flat-syndactyly foot. Results: We found a complex chromosomal rearrangement including breaks in 4q12, fragility in the 9q11-13 band region and 9qh+. Cytogenetic results agree with the literature findings. The mutation analysis of the p63 gene revealed no mutation. Conclusion: The phenotype of our patient may be due to variable expressivity and penetrance of the p63 gene and to other genetic factors, or the mutation can be located in the other 4 loci for SHFM. Additional minor modifying genes, which predispose to non-syndromic cleft palate, could also contribute to the expression of the cleft palate component of the EEC syndrome.

Key Words: Ectrodactyly, bilateral syndactyly, flat-syndactyly foot, split-syndactyly hand, p63 gene, cytogenetics

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