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A Case of Trisomy 18 Presenting with Severe Facial Abnormality and Phocomelia

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KEYWORDS Trisomy 18. Limb abnormalities. Phocomelia

ABSTRACT An unusual phenotypic presentation was found in a trisomy 18 infant, who presented facial deformity, bilateral upper limbs phocomelia and talipes equinovarus. The report added phocomelia as a phenotype of trisomy 18, which was rare in previous literature.