FRASER SYNDROME: A CASE REPORT

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INTRODUCTION: The Fraser syndrome also known as cryptophthalmia syndrome with syndactyly consists of atresia of the palpebral fissure, unilateral or bilateral often combined with eyelid and eyebrow agenesis, further malformations of the eye, ears and nose. Partial cutaneous syndactyly and other anomalies of limbs, as well as malformations of the urogenital tract are common features. Fraser syndrome is a hereditary disorder of variable expression and autosomal recessive transmission with low incidence. The prognosis depends on the severity of the malformations present and treatment are possible measures of rehabilitation. OBJECTIVE: The objective of this report is to show a multidisciplinary approach of a rare and severe case. CASE REPORT: CVVG was diagnosed with Fraser syndrome, female, four-year-old and sibling with craniosynostosis. The patient has been followed since birth by a multidisciplinary team. The child underwent facial surgeries, ventriculoperitoneal shunt further guidance with nutritionist and preventive work with speech therapists and dentists. The patient presents facial cleft, complete unilateral cleft lip and palate, left cryptophthalmia, hypertelorism, syndactyly, absence of some phalanges of fingers and toes, as well as developmental delay. Computed tomography showed semilobar holoprosencephaly, schizencephaly and hydrocephalus. CONCLUSION: This is a rare syndromic case with multiple associated deformities, which despite a poor prognosis, the development of patient is surprising and motivating for the multidisciplinary team.