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Case Report

Roberts Syndrome: A Report of a Neonatal Case

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Abstract

Roberts syndrome is a rare genetic disorder characterized by symmetrical limb malformation and craniofacial abnormalities that may be associated with organ malformations. The diagnosis is suspected clinically and confirmed by genetic testing. There is a risk of recurrence in siblings, hence the importance of genetic counseling.

We report the case of a female newborn admitted on day 1 of life with incomplete development of all four extremities and craniofacial anomalies. The clinical features were suggestive of Roberts syndrome.

Keywords: *Roberts Syndrome; Neonatal Case; Craniofacial Anomalies*

Introduction

Roberts syndrome is a rare autosomal recessive genetic disorder with diverse expression, named after John Roberts, characterized by prenatal and postnatal growth retardation with limb and craniofacial abnormalities [1,2]. The limb abnormalities are similar to thalidomide embryopathy, hence the name pseudothalidomide syndrome. Other abnormalities may be associated with this syndrome, such as cardiac and renal malformations.

Medical Observation

This is a newborn female, 2nd in a family of 2, with a healthy 4-year-old sister, from a 2nd degree consanguinity marriage. Both parents were healthy, there was no history of congenital malformation in the family, nor of abortion or exposure to teratogenic drugs during the pregnancy carried to term, without particularity.

The delivery was uneventful vaginally with an APGAR score of 8 and 9 at the first and fifth minute respectively.

The birth examination revealed a newborn with manifest facial dysmorphism consisting of incomplete development of all four limbs. Craniofacial anomalies included prominent frontal bones, prominent eyes, shallow orbits, hypertelorism, nasal hypoplasia, cleft lip and palate, low-set ears, and a short neck.

There were also limb deformities that were fixed in flexion, short with hands and feet closed to the body. The thumbs were absent bilaterally, with oligodactyly. The external genitalia were normal.

Paraclinical explorations revealed a normal chest X-ray, a cardiac and abdominopelvic ultrasound without abnormalities.

Skeletal radiographs confirmed agenesis of the radius and ulna, fusion of the 4th and 5th metacarpals with hypoplasia of the distal phalanx. The pelvic and iliac bones were normal apart from bilateral fibular aplasia.

Brain CT scan with three-dimensional bone reconstruction did not detect any parenchymal abnormality or craniosynostosis.

A constitutional karyotype revealed a newborn of female sex (46XX).

Ophthalmological exploration was normal apart from bilateral non-pulsatile exophthalmos with lagophthalmos.

In our patient, Roberts syndrome was considered due to the very telling clinical picture, genetic confirmation was not carried out due to lack of resources.

Therapeutic management was based on surgical treatment of cleft lip and palate and correction of bone malformations.

The evolution was marked by death at the age of 6 months.

Figure 1: *Showing facial abnormalities.*

Figure 2: *Hypertrophy of the lips.*

Figure 3: *View showing limb abnormalities.*

Figure 4: *Radiographic examination showing a normal pelvis*

Figure 5: *Agenesis of the radius and ulna.*

Figure 6: *Bilateral fibular aplasia.*

Discussion

Roberts syndrome is a rare autosomal recessive genetic disorder, leading to multiple congenital malformations. Features include symmetrical underdeveloped limbs with craniofacial abnormalities [3].

Limb abnormality ranges from shortening to complete absence, usually symmetrical, with short arm bones, fused fingers, and missing thumbs. The lower limbs and feet are often affected in the same way as the upper limbs.

Craniofacial malformations occur with marked variability. They include cleft lip and palate, hypertelorism, exophthalmos, a wide nasal bridge, micrognathia, and fine silver-blond hair.

Limb anomalies include tetraphocomelia which is a prominent feature of the syndrome with oligodactyly, radial aplasia or dysplasia.

Other malformations associated with this syndrome include cardiac anomalies: atrial septal defect, ventricular septal defect, and patent ductus arteriosus. They also have polycystic or horseshoe kidneys.

Genital anomalies such as penile hypertrophy, cryptorchidism, clitoral hypertrophy. Neurological anomalies such as microcephaly, hydrocephalus and tibial femoral ankylosis [3].

Intellectual disability is often present in most cases. Some may have normal intelligence.

The diagnosis of Roberts syndrome can be made clinically in children with characteristic intrauterine growth retardation, characteristic limb malformations, and craniofacial anomalies. However, the gold standard for diagnosis remains cytogenetics by screening for $ESCO_2$ spectrum disorders [4].

Several syndromes pose the differential diagnosis with Roberts syndrome but two syndromes are clinically closer, namely pseudothalidomide and TAR (thrombocytopenia without ray syndrome).

Roberts syndrome can be suspected prenatally by the observation of characteristic abnormalities on antenatal ultrasound and confirmed by cytogenetic testing [1,5].

To assess the risk of recurrence, molecular genetic testing is recommended for parents to confirm that each is heterozygous for a pathogenic $ESCO_2$ variant.

In our case, the diagnosis was made based on clinical characteristics at birth, cytogenetic tests were not performed due to the high cost.

Treatment of mild forms aims to improve the quality of life of children with this syndrome. Possible corrections include cleft lip and palate correction, surgical correction of limb abnormalities and management of cognitive impairment, nutritional rehabilitation, and parental counseling [6,7].

The prognosis depends on the severity, degree of malformation and involvement of other organs [6].

Conclusion

Roberts syndrome is rare, a diagnosis with precise genetic counseling allows the risk of recurrence to be assessed, and management is multidisciplinary.

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