

SPOT DIAGNOSIS (IMAGE GALLERY)



HYPOPLASTIC LIMBS AND CLEFT LIP, PALATE

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A female baby with weight of 2.4 kg was born to a primigravida 18 years old mother. Baby was a product of non-consanguineous marriage and delivered at 36 weeks of gestation by normal vaginal delivery. Mother's gestational period was uneventful with no history of teratogenic drug intake or radiation exposure. Baby had hypoplastic forearm along with flexion contractures at wrist joint. She had aplasia of both lower limbs and feet were attached directly to the trunk. In addition she was found to have oligodactyly in right hand, cleft lip and cleft palate.

What is the likely diagnosis?

Roberts syndrome (RBS) and SC phocomelia are autosomal recessive disorders. (1) RBS is characterized by symmetric limb reduction defects, craniofacial anomalies (skull, eyes, lip, and palate), growth retardation, cardiac and renal abnormalities, flexion contractures of various joints and varied intellectual deficit. (2) These two syndromes represent part of a spectrum, with RBS at the most severe range in which severely affected infants may be stillborn or die in the post-natal period, while individuals with SC phocomelia correspond to the milder end of the spectrum and typically survive to adulthood. (3) Upper limb phocomelia is more common than lower limb phocomelia. Aplastic or hypoplastic thumbs, oligodactyly or syndactyly can also occur. Differential diagnosis include DK Phocomelia syndrome, Odontotrichomelic tetramelic ectodermal dysplasia, congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD) syndrome, syndrome of spleno-gonadal fusion. (4-8) RBS is also termed as "pseudo-thalidomide" syndrome because phenotypically it shares similarities to those caused by the ingestion of thalidomide by pregnant woman. (9) When a previous child in the family is diagnosed with RBS and carries ESCO2 mutations, prenatal diagnosis can be offered. Cytogenetic analysis of fetal cells obtained from chorionic villi samples at the first trimester, amniocentesis, or cord centesis during the second and third trimesters, is required to confirm the diagnosis prenatally. RBS may be suspected by observation of characteristic RBS anomalies at ultrasonography. Most individuals with RBS are stillborn or die in infancy when detected before viability, termination of pregnancy can be offered. (10) Management includes surgical correction of facial malformations, surgical and, or orthopedic treatment of limb defects and management of the cognitive disabilities. High mortality in the newborn period or early childhood is due to cardiac or renal malformations.

References

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