

# Prenatal Diagnosis of Roberts-SC Phocomelia Syndrome: Case Report

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## Abstract

**Background:** Roberts-SC phocomelia syndrome (RS) is an autosomal recessive disorder. We report here a case in which prenatal pathologic sonographic, cytogenetic premature centromere separation (PCS) findings were suggestive of this syndrome. The findings were in correlation with the postmortem examination.

**Case:** Our patient is 38 years old, G2P0 who has consanguineous marriage. Cleft palate/lip, severe upper and lower limb deficiencies and bilateral 4 fingers of hands were detected in the ultrasound scan conducted at 18 weeks of gestation. Cytogenetic prenatal analysis was carried out due to advanced maternal age and pathologic findings in the ultrasound which led to the finding of premature centromere separation (PCS). Postmortem examination of the fetus was done after termination which confirmed the sonographic findings as well as hypertelorism, bilateral cleft lip and cleft palate, tetraphocomelia and penile enlargement, bilateral pes equinovarus.

**Conclusion:** This case emphasized the importance of the sonographic and PCS findings during prenatal analysis of Roberts SC syndrome as well as the importance of the postmortem examination for the confirmation of prenatal diagnosis.

**Keywords:** The Roberts-SC phocomelia syndrome, prenatal diagnosis, premature centromere separation.

## Roberts- SC fokomeli sendromunun prenatal tanısı: olgu sunumu

**Amaç:** Robert-SC Fakomeli, otosomal resesif geçişli nadir bir hastalıktır. Bu çalışmada patolojik ultrason ve fetal sitogenetik 'erken sentromer ayrılması' bulguları nedeniyle Roberts-SC sendromu olduğu düşünülen ve postmortem muayene sonuçlarıyla da bu tanı ile uyumlu olduğu görülen bir olgunun sunulması amaçlanmıştır.

**Olgu:** 38 yaşında, G2P0, ilk gebeliği abortus ile sonuçlanan anne adayının ikinci gebeliğinin 18.haftasında gerçekleştirilen ultrasonografik inceleme sonucunda fetüste yankı damak/dudak, alt ve üst ekstremité kısalığı, bilateral 4 el parmağı gibi bulgular izlendi. İleri anne yaşı ve patolojik ultrasonografik bulgular nedeniyle fetal kromozom analizi yapılmıştır. Bu analiz sonucunda özellikle akrosentrik kromozomlarda 'erken sentromer ayrılması' (PCS) bulgusu gözlemlendi. Gebeliğin terminasyonu sonrası gerçekleştirilen postmortem muayene sonucunda fetusun belirgin hipertelorizm, bilateral damak/dudak yarığı, tetrafakomeli, belirgin penis, bilateral pesekinovarus deformitesi gibi bu sendrom ile uyumlu klinik bulgularının olduğu gözlemlendi.

**Sonuç:** Bu olgu ile Roberts-SC Fokomeli sendromunun prenatal tanısında fetal ultrasonografik inceleme sonrası elde edilen bulguların yanı sıra fetal sitogenetik incelemedeki "erken sentromer ayrılması" bulgusuna dikkat edilmesi gerektiği ve postmortem muayene ile de tanının doğrulanmasının önemi vurgulanmıştır.

**Anahtar Sözcükler:** Roberts-SC fokomeli sendromu, prenatal tanı, erken sentromer ayrılması.

## Background

Roberts-SC phocomelia syndrome is an autosomal recessive disorder in which many craniofacial anomalies and various extremity defects exist.<sup>1</sup> It is characterized with prenatal and postnatal growth retardation and death is observed in dead birth and early infancy periods of many cases.<sup>2</sup> Extremity defects in this syndrome may change from hypomelia to tetraphocomelia and cleft palate-lip, midfacial hypoplasia, micrognathia, prominent eyes and hypertelorism are deemed as craniofacial anomalies.<sup>3</sup>

In many Roberts-SC phocomelia cases, 'Premature Centromere Separation' (PCS) diagnosis is found cytogenetically.<sup>4</sup> By this case, it is aimed to present prenatal ultrasonographic and cytogenetic diagnoses of rarely observed Roberts-SC phocomelia syndrome and also postnatal examination diagnoses.

## Case

38 years-old, G2P0, prospective mother who had a miscarriage in her first gestation, who is married to her cousin in the first remove did not have any feature in her family background. Microcephaly, cleft palate-lip, lower and upper extremity shortness (tetraphocomelia), bilateral 4 fingers in hands were observed in fetus on advanced ultrasonographic examination performed on 18<sup>th</sup> week of second gestation. Fetal chromosome analysis was suggested to prospective mother by giving genetic consultation due to advanced maternal age and pathological ultrasonography diagnoses. After amniocentesis process applied to prospective mother, prometaphase chromosomes obtained from amniotic fluid culture were examined by using GTG banding technique. As a result of analysis, PCS diagnosis was found in acrocentric chromosomes especially in all metaphase areas (Figure 1).

It was thought by these ultrasonographic and cytogenetic diagnoses that fetus had Roberts-SC phocomelia syndrome. The fetus was discharged by the demand of the family after giving genetic consultation to them.

After postmortem examination, crown-rump length of 280 gr male fetus was measured as 15 cm



**Figure 1.** Diagnosis of premature centromere separation observed in acrocentric chromosomes in fetal chromosome analysis.

and crown-heel length was measured as 17.5 cm. There were dysmorphic diagnoses in crown neck area of the fetus such as short neck, microcephaly, dysplasia and lower located ears, hemangioma in glabellas, hypertelorism, prominent eyes, midfacial hypoplasia, small back located chin, bilateral cleft palate/lip. Tetraphocomelia, bilateral 4 fingers in hands, bilateral pes equinovarus deformity were observed in extremities. Also, there were diagnoses such as hypoplastic pelvis and penile clarity (Figure 2).

## Discussion

Roberts-SC phocomelia syndrome is a kind of rarely seen autosomal recessive disorder which includes various clinical diagnoses. Pre/postnatal growth retardation and mental retardation are seen in this syndrome together with extremity and craniofacial anomalies. Diagnoses such as tetraphocomelia, flexion contractures, cleft palate/lip, hypoplastic alar nasi, prominent eyes, hypertelorism and facial hemangioma are deemed within these anomalies and most of the cases are lost.<sup>5,6</sup> Roberts-SC phocomelia syndrome can be recognized by ultrasonographic and cytogenetic examinations performed during prenatal period.<sup>7,8</sup> Intrauterine growth retardation, microcephaly, lower and upper extremity shortness and cleft palate were observed in



**Figure 2.** Frontal view of fetus in which diagnoses are observed in postmortem examination such as cleft palate/lip, tetraphocomelia, clear penile.

our case as a result of ultrasonographic examination performed in 18<sup>th</sup> gestational week.

While PCS diagnosis was found in many of the cases with Roberts-SC phocomelia syndrome as a characteristic chromosomal diagnosis, some cases with normal chromosomes were also reported.<sup>9</sup> As a result of fetal chromosome analysis performed in amniotic fluid culture of our case, PCS diagnosis was observed especially in acrocentric chromosomes. Pathological ultrasonographic and cytogenetic diagnoses are very important in prenatal diagnosis of this syndrome. But confirming this diagnosis as postnatal has an important role in terms of the way to be followed during and after giving consultation to families. After postmortem examination for our case; clinical diagnoses compatible with Roberts-SC phocomelia syndrome

such as prominent eyes, hypertelorism, hemangioma in glabellas, midfacial hypoplasia, dysplasia and lower located ears, pelvic hypoplasia and penile clarity were observed in fetus as well as ultrasonographic diagnoses and it was confirmed as postnatal.

## Conclusion

Fetal ultrasonographic cleft palate/lip and extremity defects in prenatal diagnosis of Roberts-SC phocomelia syndrome and "premature centromere separation" diagnosis in cytogenetic examinations should be especially paid attention. Furthermore, this diagnosis should be confirmed by postmortem examination. With the help of all this information, giving genetic consultation about disorder clinic, prognosis and repetition risks to the family will provide appropriate observation and process possibilities for other prospective pregnancies.

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