Short-rib polydactyly syndrome: antenatal diagnosis

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Objective
Presentation of antenatal diagnosis of short-rib polydactyly syndrome. Short-rib polydactyly syndromes are rarely encountered and consist of four subtypes. All have micromelia, thoracic hypoplasia and polydactyly in common. These are inherited in an autosomal recessive pattern. Prognosis of these cases is invariably lethal.

Methods
A case report of short-rib polydactyly syndrome.

Results
A 21 year-old patient at 17 weeks of gestation (gravida 2, previous pregnancy was terminated with the diagnosis of skeletal dysplasia) was referred to our department because of poor obstetric history. During sonogram, thoracic hypoplasia with short ribs, atrioventricular septal defect, severe micromelia and postaxial polydactyly were detected. The parents were counselled about the condition and they opted for termination of pregnancy. Sonographic diagnoses were confirmed after termination. In regard to the patient's previous obstetric history, sonographic and post-mortem findings, one of the subtypes of short-rib polydactyly syndromes was diagnosed.

Conclusion
The exact incidence of the short-rib polydactyly syndromes is not known. Prognosis is extremely poor, so patients should be counselled on the option of termination of pregnancy. Because of the autosomal recessive inheritance pattern, the recurrence risk is 25%. Those cases with a history of short-rib polydactyly syndrome should be counselled on early screening in their future pregnancies.