A case of multiple pterygium syndrome
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Objective
Multiple pterygium syndrome consists of a wide range of fetal malformations which have a genetic link. There are two types of multiple pterygium syndrome (MPS): lethal and nonlethal. The nonlethal form of MPS is called Escobar syndrome. The mode of inheritance can be either autosomal recessive, autosomal dominant, or X-linked dominant. We present a case of lethal MPS presented at our institute.

Methods
This is a case report.

Results
A 27-year-old G4P1A2L1 patient was referred to our clinic for detected cystic hygroma at 11 gestational weeks. She had 2 previous pregnancy loss in the second trimester because of hydrops fetalis with normal karyotype. The patient had a nonconsanguineous marriage. Ultrasound was performed at 12 gestational weeks. The fetal CRL was 58 mm and was compatible with 12 weeks and 2 days. The two-dimensional (2D) ultrasound showed a cystic hygroma (10.9mm), a reversed flow of ductus venosus, joint contractures and fetal akinesia. Fetal karyotyping was done by CVS (chorion villus sampling). Conventional karyotyping was normal. After two weeks, control sonography showed fetal hydrops and the contracture in the extremities. At family request, the pregnancy was terminated at 15 gestational weeks. After the termination of pregnancy, the fetus was morphologically examined and multiple pterygia involving fingers, bilateral antecubital, popliteal, and intercrural area were found. We plan to carry on advanced genetic testing of fetus.

Conclusion
Multiple pterygium syndrome is a rare disease, and it may be explanation for recurrent cystic hygroma. Genetic counseling is essential for future pregnancies as both lethal and Escobar variant of MPS has autosomal/X-linked inheritance.