PRENATAL DIAGNOSIS OF ECTRODACTYLY BY ULTRASONOGRAPHY

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OBJECTIVE
Ectrodactyly is an ectodermal dysplasia affecting the limbs, causing absence of central digits with a median cleft and fusion of the remaining lateral digits. We present a case of early diagnosis using ultrasound.

METHODS
A 20 year-old nulliparous woman, presented at 12.3 weeks of gestational age for first-trimester screening for aneuploidy. The patient’s husband had bilateral hand malformation, showing lobster-claw like deformity. There was a consanguinity history between both. On 2D imaging it could be noted that upper limbs presented decreased movements and abnormal position, with pincer shape. Nuchal translucency was within normal limits, measuring 1,08mm. Chorionic villus sampling was suggested, but even though extensive counseling, the couple rejected it, understanding and accepting the possible consequences of this fetal malformation. On subsequent imaging, both humeri showed normal length, but forearms were shortened. Hands remained in hyperflexion and only two fingers could be visualized on the left hand, and three on the right. Not other anatomic defects were detected.

RESULTS
The term ectrodactyly comes from the Greek words ektroma (abortion) and daktylos (finger). It is a rare autosomal dominant ectodermal dysplasia, which may sometimes be associated with other ectodermal defects such as cleft lip/palate and tear duct anomalies. This malformation is due to abnormal development of the hand/foot plates during de seventh week of pregnancy. Normally, five digital rays arise from de hand and foot plates, and the apical ectodermal ridge leads the growth and differentiation of each ray. Central rays form de second, third and fourth digits. In ectrodactyly this ray is abnormal, and apical ectodermal ridge ceases to function correctly.

CONCLUSION
Early diagnosis of fetal limb abnormalities may be important because those findings can be associated with other anomalies. This will provide more time for patients counseling and decision making about terminating or continuing de gestation.