A rare case of distal phocomelia: a case report
A comparison between prenatal ultrasound findings, fetal autopsy and X-Ray

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Introduction
Phocomelia is a rare developmental anomaly during pregnancy. It’s a lack of development of limbs (one or more).
We present a case of a termination of pregnancy for a rare case of phocomelia diagnosed by prenatal ultrasound

Objective
Insist on the importance of the morphological first trimester ultrasound in detecting limb abnormalities. Discuss the pathogenesis of phocomelia.

Case report
We present the case of a patient aged 35 years, blood group O positive, diabetic on Insulin since the young age. She has been married for 10 years, non consanguineous marriage, G III P II, 2 children living in apparent good health. Current pregnancy not initially followed, the first trimester ultrasound was not performed, triple test not done. The patient was referred to our department after the discovery during the 1st fetal ultrasound (23 weeks of gestation) of an abnormality in the upper limbs: (fig 1,2) agenesis of the forearms and bilateral clubfeet..

The karyotype was normal 46 XX

Discussion
Phocomelia is stopping development of one or more members, it is characterized by atrophy of the limbs leading to somehow direct implantation of the hands and feet directly on the trunk. There are three types of phocomelia:
• Full phocomelia in which the hand and foot are attached directly to the trunk.
• The proximal phocomelia in which the abnormality concerns the arm and thigh.
• The distal phocomelia with aplasia (lack of development) of the forearm or leg.
Many cases of phocomelia were caused by the prescription of thalidomide, a sedative marketed in 1960 with one of the enantiomers proved toxic to the child. Since the end of commercialization of this product, we noticed the decrease in cases of phocomelia.
This defect is usually not isolated and associated with a chromosomal abnormality.
Studies suggest that diabetes poorly controlled can be a risk factor.
The first trimester ultrasound is useful for early diagnosis and management.

Conclusion
Phocomelia is a rare sporadic malformation.
Morphological first trimester ultrasound has a major importance in the early diagnosis and subsequent management.