



A case of Fraser Syndrome

Azevedo IM, Martins I, Rodrigues AC, Nunes F, Nascimento C
Hospital de Cascais, Lisboa, Portugal

Objective

To analyse and compare the characteristics and morphology of a fetus with Fraser syndrome diagnosed in first trimester ultrasound.

Methods

Review of the literature about Fraser Syndrome. Analysis of the morphology of the fetus on ultrasound, radiography and pathology. Scientific drawing.

Results

Minor criteria for the disease are the multiple skeletal malformations of the fetuses being analysed, which are observed on ultrasound. The major criteria -cutaneous syndactyly, abnormalities of the genitalia and microphthalmia, could only be identified after medical abortion.

Conclusion

Cryptophthalmos is the most common and specific abnormality in people with Fraser syndrome, however this is very difficult to identify during first trimester ultrasound. The ultrasound can identify a fetus with poly-malformative syndrome. The final diagnosis is made by pathological examination.



