First trimester diagnosis of frontonasal dysplasia

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Objective

Presentation of intrauterine diagnosis of frontonasal dysplasia. Frontonasal dysplasia is a rare malformation characterized by hypertelorism, anterior cephalocele, median cleft lip/palate and bifid nose. Prognosis depends on the severity of the defects. Prenatal diagnosis in early pregnancy is very rare, to the best of our knowledge.

Methods

A case report of frontonasal dysplasia.

Results

A 23 year-old patient at 13 weeks of gestation (gravida 1 para 0) was admitted to our department for first trimester aneuploidy screening. During sonogram, anterior cephalocele, hypertelorism and median cleft involving lip and nose were detected. The parents were counselled on the condition and they opted for termination of pregnancy. Sonographic diagnoses were confirmed after termination. Considering sonographic and postmortem findings, frontonasal dysplasia was diagnosed.

Conclusion

The incidence of frontonasal dysplasia is not known. To date, only case reports associated with antenatal diagnosis have been published. The Inheritance pattern has not yet been defined. However, genetic counselling is important regarding positive family history. Frontonasal dysplasia should be considered in differential diagnosis of cases with anterior cephalocele.