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A case of Antley-Bixler syndrome

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Objective

Antley-Bixler syndrome (ABS) is a rare syndrome characterized by abnormal fusions of certain bones, particularly the skull, hips and arms. The karyotype is normal. The identification of ABS from bone malformations in prenatal sonography is underestimated: 40 cases listed, 3 published cases of prenatal diagnosis.

Methods

We report a case of a Antley-Bixler syndrome at the Department of Obstetrics & Gynecology, at Charles Nicolle Hospital of Tunis, Tunisia.

Results

We present a case of a 31 years old patient with no previous medical history, currently in her second ongoing pregnancy who presented at 24 weeks due to amenorrhea. A complete medical examination was performed without abnormal findings. An ultrasound examination on the fetus reported: a hypoplastic skull in the medial part of the face with lacunar aspect, curved long bones and radio-humeral synostosis. No invasive testing was done and the couple opted to terminate the pregnancy. Pathological examination of the fetus showed a craniofacial dysmorphia, macrocrania, large anterior fontanel, a meningocele, hypertelorism with exophthalmos, low implanted ears, crushed nose, thin lips with half-open mouth, microretrognathia, wide neck with excess skin, brachycephaly, median palmar crease, limitation of the extension of the limbs, bilateral piolet feet, atrial septal defect and incomplete lobulation of the right lung. The treatment of Antley-Bixler syndrome is symptomatic, with notably early neurosurgical and pneumological management. The prognosis remains poor, with the majority of patients dying in childhood due to respiratory complications.

Conclusion

Antley-Bixler syndrome is a rare autosomal recessive genetic disease characterized by a high number of bone malformations. The antenatal diagnosis is based on the ultrasound findings.