Published in final edited form as:

Am J Dis Child. 1991 June ; 145(6): 701–702. doi:10.1001/archpedi.1991.02160060119033.

Picture of the Month

Merlin G. Butler, MD, PhD, Sarah Hassell, MD [Contributors], Murray Feingold, MD [Editor for This Case], and Walter W. Tunnessen Jr, MD [Section Editor]

Department of Pediatrics, Division of Genetics, Vanderbilt School of Medicine, Nashville, Tenn

Denouement and Discussion

Antley-Bixler Syndrome

Manifestations—Major manifestations include craniosynostosis, a characteristic facial appearance, choanal atresia/stenosis, radiohumeral synostosis, bowing of femurs with fractures, and multiple joint contractures.

Craniofacial abnormalities consist of brachycephaly secondary to craniosynostosis of the coronal and lambdoidal sutures; large anterior fontanelle; and a face characterized by frontal bossing, flat facies, severe midface hypoplasia, proptosis, flattened nasal bridge, pear-shaped nose, choanal stenosis/atresia, long philtrum, and small, dysplastic ears.

Skeletal abnormalities include radiohumeral synostosis, anterior bowing of femurs with neonatal fractures, medial bowing of ulnas, narrow iliac wings, arachnodactyly with distal tapering of digits, camptodactyly, and scoliosis. Other occasional abnormalities are hydrocephalus, atrial septal defects, vaginal atresia, renal defects, multiple hemangiomas, and preauricular tags. Auditory dysfunction may also occur.

In most reported cases, prenatal and postnatal growth have been normal. Respiratory compromise generally occurs secondary to upper-airway obstruction (choanal stenosis/atresia occurs in about one half of affected individuals) and may vary from nasal congestion to apnea leading to death. Some patients have died in infancy of unexplained causes. Patients followed up beyond age 1 year have been ambulatory. Intelligence varies from normal to mild retardation.

Genetics—Antley-Bixler syndrome occurs sporadically, but has been reported in siblings and children of consanguineous parents. Therefore, inheritance is most likely autosomal recessive with a 25% recurrence risk for the condition in subsequent children from the same parents.

Treatment—Treatment consists of management of the orthopedic abnormalities, including the multiple joint contractures, through physical therapy, casting, and/or surgery. Surgery may also be necessary to alleviate upper-airway obstruction and to correct the

craniosynostosis. Hearing, cardiac, and renal evaluations are also indicated. Genetic counseling and prenatal diagnosis should be discussed with the family.

References

1. Antley RM, Bixler D. Trapezoidocephaly, midface hypoplasia and cartilage abnormalities with multiple synostoses and skeletal fractures. *Birth Defects Original Articles Series*. 1975; 11:397–401.
2. DeLozier CD, Antley RM, Williams R. The syndrome of multisynostotic osteodysgenesis with long-bone fractures. *Am J Med Genet*. 1980; 7:391–403. [PubMed: 7468663]
3. Escobar LF, Bixler D, Sadove M, Bull MJ. Antley-Bixler syndrome from a prognostic perspective: report of a case and review of the literature. *Am J Med Genet*. 1988; 29:829–836. [PubMed: 3041834]

Figure 1.

Figure 2.

Figs 1 and 2.

3-week-old newborn with brachycephaly; prominent forehead; flat face; low-set, dysplastic ears; flattened nasal bridge; pear-shaped nose; severe, midface hypoplasia; proptosis; flexed position of elbow; and camptodactyly.

Fig 3.
Radiohumeral synostosis.

Fig 4.
Bowling of the femur.