Antley-Bixler Syndrome In A Nigerian Child
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Citation

Abstract
Antley-Bixler syndrome is a rare multiple congenital anomaly with many musculoskeletal abnormalities and high mortality rate. We present the first Nigerian 5-year old girl from unrelated parents with bilateral humeroscapular and humeroribs synostosis, humeroulnar synostosis, acetabulofemoral synostosis, midface hypoplasia, craniosynostosis, arachnodactyly, large clitoris, and multiple joint contractures. Surgical excision of bone bars (bilateral Shoulder, left elbow) and release of joint contractures was performed. She is still being followed-up at Orthopedics clinic 2 years post operative with improved functional capacity. About 43 such cases had been reported in the past (1).

CASE PRESENTATION
A 5-year old Nigerian, born to a 37 year father and 32 year old mother was referred to the Orthopedics outpatient clinic of Wesley Guild Hospital, Ilesa with difficulty in hip abduction for perineum care.

Pregnancy was full term and uneventful. No history of fluconazole ingestion by mother during pregnancy. Delivery was by spontaneous vaginal vertex presentation. She began to walk at the age of one year and was fully immunized. The family history was not contributory. She is the second child of the parent’s three children and the other siblings are normal.

She developed spontaneous bony swellings on the head and the body. The bone bar on the right arm and shoulder were first noticed at the age of one year. She had difficulty in sitting up from a supine position and falls down easily on walking. She however can sit down from erect posture easily. The abnormal bony prominences in the back and stiffness of the joints increased progressively. The right hand could just reach her mouth and the left cannot. Intellectual capacity is normal for age although yet to commence formal education due to her deformities.

Examination revealed a moderately active young girl having cephalous-trapezoidal head with frontal bossing, midface hypoplasia, and craniosynostosis (multiple nodules of bony growth mainly in occipital region). Thoracolumbar scoliosis with convex to the right (cob angle < 30°). No spinal dysraphism. The limbs examination showed prominent scapulae, bilateral shoulder ankylosis and global atrophic muscles of the upper limbs. The shoulder was ankylosed at 45° abduction. Elbows were held ankylosed in full extension. The wrist joints are freely mobile. The ankle, foot and knee joints are normal. Bilateral gluteal muscles atrophy, limited passive hip flexion and extension. She has multiple joint contractures at the shoulders, elbows, hips and arachnodactyly fingers. No bowing of femur, tibia, ulna and radius. The abdominal and genitourinary examination showed hepatomegaly, hypo plastic labia major and large clitoris.

Investigations revealed normal hematological and biochemical results. The plain radiographs in Figures 1-4, show bilateral humeroscapular and humeroribs synostosis, left humeral ulna synostosis, acetabulofemoral synostosis, and thoracic-lumbar scoliosis. Abdominal ultrasound confirmed hepatomegaly. Echocardiography shows sinus arrhythmias and ectopic beats.

The surgeries performed were excision of bony bars of humeroscapular, humeroradius synostosis and soft tissue contractures release. Genetic counseling was provided to her parents. Wounds healed and there was improvement of gait and functional capability including walking, eating and shoulder mobility. She has been followed-up for two years without complication.
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Figure 1
Figure 1ab: Bilateral humeroscapular and humeroribs synostosis

Figure 2
DISCUSSION

Antley-Bixler Syndromes (ABS) is a rare disorder characterized by multiple malformations of cartilage and
bone including multisynostotic osteodysgenesis first reported in 1975. The aetiopathogenesis of ABS is still unknown (1). It has not been reported in Nigeria or indeed Africa.

The 5-year old Nigerian girl presented with clinical and radiological features of ABS including craniosynostosis, bilateral humerocapular and humeroribs synostosis, midface hypoplasia, left humeroulnar synostosis, multiple joints contractures which satisfied the clinical hallmarks (2). She represented sporadic occurrence in the otherwise normal family (3). The mode of inheritance is probably autosomal recessive (4) but there is no racial predilection. A female preponderance has been reported (male: female ratio of 2:7) (5). The hypoplastic labia major and large clitoris are urogenital anomalies seen in less than 50% of ABS. The ectopic beats reflected underlying congenital heart defect noted in 21% of cases (6).

The involvement of humeroribs and acetabulofemoral synostosis is unique in our patient. Humero radial synostosis was present in almost all cases reported, few have humeroribs and acetabulofemoral synostosis separately. The survival of the patient is related to the absence of severe respiratory and urogenital anomalies and prognosis improves with age (2). The parents are not related but consanguinity has been reported twice in ABS (7).

Mid trimester ultrasound has been found useful in prenatal diagnosis of ABS (8). Fluconazole taken in the first trimester is associated with increased risk of ABS (9). Bone synostosis frequently results in early orthopedic consult and the surgeons should be aware of the frequent associations with ABS. A high index of suspicion is necessary for diagnosis. In all cases of neonatal death from severe respiratory distress, examination of the joints and if necessary postmortem CT scan and X-Ray may help decrease number of cases missed.

Early intervention addressing major congenital malformations form the mainstay of treatment (3). The surgical excision of the bilateral shoulder and elbow bone bars with soft tissue contracture release has improved the functional capacity of the patient. The two years follow-up post surgery is satisfactory.

CONCLUSION

Antley-Bixler Syndrome diagnosed in the 5-year old Nigerian girl presented classically and uniquely with radio humeral, humeroribs and acetabulofemoral synostosis. Awareness of the ABS by Orthopedic Surgeons will improve early diagnosis, treatment, and prognosis in Orthopedic practice.

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References

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