

INTRODUCTION

Antley-Bixler syndrome (ABS) was first described by Ray Antley and David Bixler in 1975. The syndrome is rare and characterized by abnormalities of bone and cartilage development (“syndrome of multisynostotic osteogenesis”). Craniosynostosis and radiohumeral synostosis are the most consistent findings. The cause of this syndrome is unclear, but genetic mutations are suspected.

CASE REPORT

A 21-year-old female G2P1A0 was referred to the obstetrics and gynecology outpatient clinic at Bhakti Husada Hospital at 32-33 weeks of gestation for antenatal care and further evaluation due to suspected fetal craniosynostosis and ventriculomegaly. At 38 weeks’ gestation, the baby was born with craniosynostosis, ocular hypertelorism, and syndactily digiti II-IV pedis dextra and sinistra, suspected Apert syndrome. Further investigation by using radiologic examination (cranial MSCT scan and X-ray at anteroposterior and lateral articulation cubital) revealed craniosynostosis, bilateral radioulna and radiohumeral synostosis, directed the diagnosis to Antley-Bixler syndrome. The baby was discharged after 21 days of hospitalization. Currently, the baby is still undergoing regular evaluation and treatment at the Ophthalmology and Neurosurgery outpatient departments at Kariadi General Hospital.

CONCLUSION

Antley-Bixler syndrome is a rare syndrome. It is very similar to Apert syndrome but can be differentiate after the baby was born and do the x ray examination it shown bilateral radiohumeral and radioulnar synostosis. The prognosis on this case depends on the progression further management craniosynostosis and ventriculomegaly.



Figure 1. Transabdominal ultrasonography shown cloverleaf skull (craniosynostosis), ventriculomegaly unilateral, exorbitismus, dan syndactily



Figure 2. General Examination Neonatal shown that craniosynostosis, ocular hypertelorism, disertai syndactily digiti II-IV pedis dextra et sinistra with early diagnose Apert Syndrome.



Figure 3. X-rays of the anteroposterior and lateral cubital joints show fusion of the radius and ulna bilaterally, as well as bilateral fusion of the radius and humerus with bilateral radioulnar synostosis and bilateral radiohumeral synostosis seen on anteroposterior and lateral cubital joint radiographs.

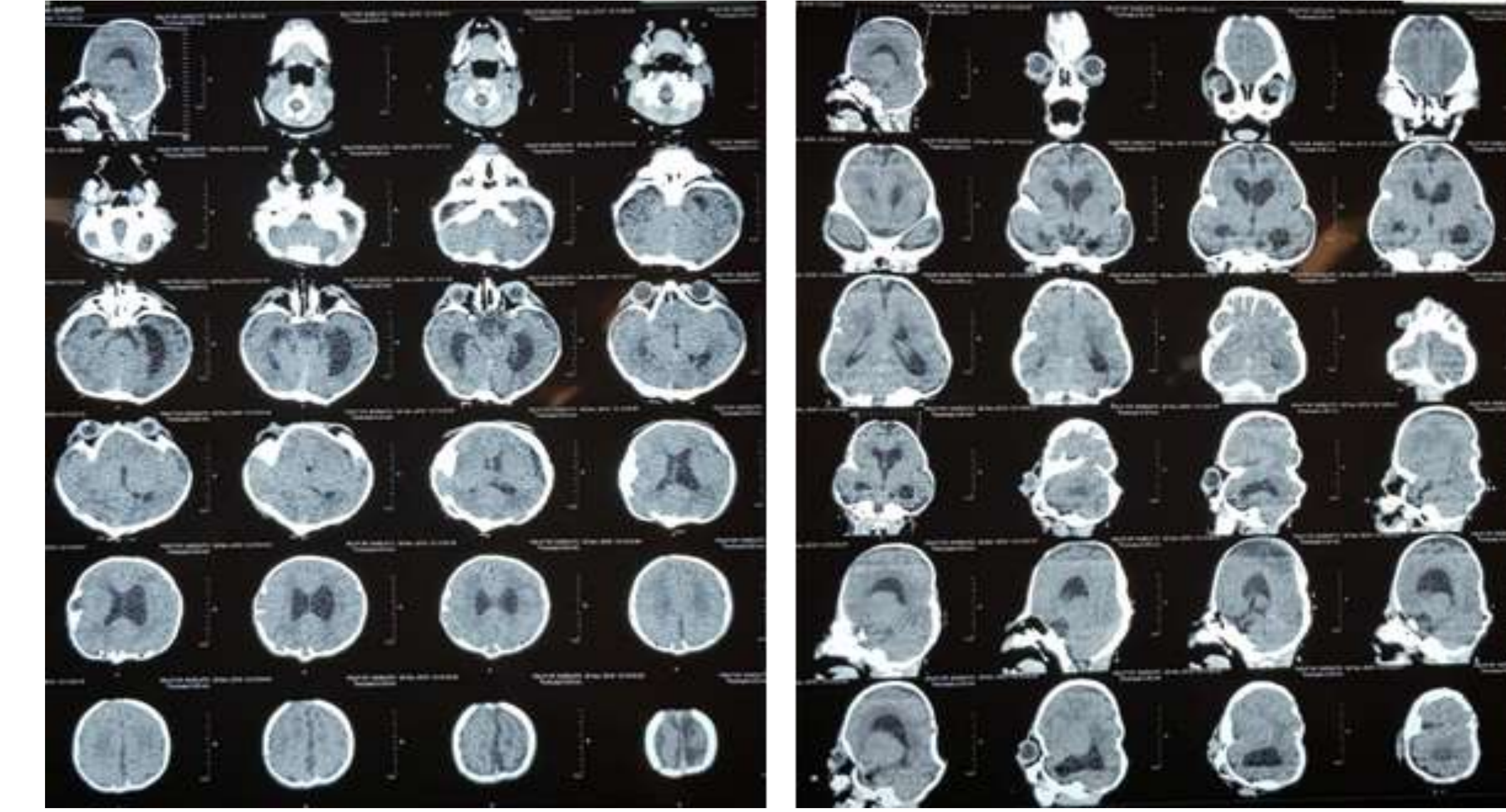


Figure 4. Non contrast cranial MSCT Scan showed turriccephaly type craniosynostosis with multiple areas of non ossification on the calvarial bone and bone spiculation on the cerebral sulci, obstructive hydrocephalus with obstruction of the foramen of Monro accompanied by transependymal edema, small posterior fossa and bilateral proptosis oculi with hypertelorism

DISCUSSION

In this case report at early examination USG lead to Apert Syndrome because found craniosynostosis & syndactily digiti II-IV pedis dextra and sinistra, but further evaluation founded synostosis radiohumeral and radioulna bilateral so we diagnose to Antley Bixler Syndrome. ABS is a syndrome that affect the develop bone & cartilage as known as multisynostotic osteodysgenesis syndrome. There two types of ABS ; ABS type I is autosomal dominan which affect FGFR gene not affect steroidogenesis, ABS type II is autosomal resesive which mutation POR gene affect deficiency glucocorticoid (congenital adrenal hyperplasia). Some of them also said exposure to high dose of fluconazole can contribute this (inhibit cP450), which is enzyme for synthesis adrenal hormon & steroid gonadal. Morbidity & mortality rate for this disease depend on what congenital acquired. Existing stenosis/atresia choana can obstruct upper airway with respiratory disturbance like recurrent infection. Correction for craniosynostosis do at age 3-6 months, and also radiohumeral synostosis.

REFERENCES

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