Antley-Bixler syndrome

Genetics

-Gene: POR (NADPH-cytochrome P450 reductase; 7p11.2)

-AR

Clinical findings/Dysmorphic features

-Ambiguous genitalia, enlarged cystic ovaries, poor masculinization in males, maternal virilization during pregnancy with an affected fetus

-Craniosynostosis, choanal stenosis/atresia (nose blockage), stenotic external auditory canals, hydrocephalus

-Neonatal fractures, bowing of the long bones, joint contracture, renal malformations

Etiology

-Only 140 cases reported

Pathogenesis

-Disorder of steroid and cholesterol synthesis due to cytochrome P450 reductase deficiency

Genetic testing/diagnosis

-Sterol or steroid abnormalities using GC-MS

-Increased urinary pregnenolone and progesterone metabolites

-POR sequencing (92%); In/Del analysis (2.5%)