Gorlin-Chaudhry-Moss syndrome is an extremely rare inherited disorder characterized by premature closure of the fibrous joints (sutures) between certain bones in the skull (craniosynostosis), unusually small eyes (microphthalmia), absence of some teeth (hypodontia), and/or excessive amounts of hair (hypertrichosis) on most areas of the body. Affected individuals may also exhibit a mild delay in physical development (growth retardation); short fingers and/or toes; and/or underdevelopment (hypoplasia) of the two long folds of skin on either side of the vaginal opening (labia majora) in females. In addition, there may be an abnormal opening between the two large blood vessels that carry blood away from the heart (pulmonary artery and aorta), causing inappropriate recirculation of some blood through the lungs, rather than throughout the rest of the body (patent ductus arteriosus). In some cases, mild mental retardation may also be present. It is believed that Gorlin-Chaudhry-Moss syndrome may be inherited as an autosomal recessive trait.
Gorlin-Chaudhry-Moss syndrome is an extremely rare inherited disorder that is apparent at birth (congenital). Approximately four cases have been reported in the medical literature. Although all reported cases have involved females, the true ratio of affected females to males is not known. The first case of Gorlin-Chaudhry-Moss syndrome was reported in the medical literature in 1960. Gorlin-Chaudhry-Moss syndrome may be diagnosed at birth, based upon a thorough clinical evaluation and characteristic physical findings. The presence of patent ductus arteriosus occurring in association with Gorlin-Chaudhry-Moss syndrome may be determined by a variety of tests. When an abnormal heart murmur is detected, chest X-rays may be ordered along with an electrocardiogram (ECG), a test that measures the heart muscle's electrical activity. Structural and functional abnormalities of the heart and its blood vessels can be analyzed through the reflection of sound waves (echocardiogram). Conductive hearing loss occurring in association with Gorlin-Chaudhry-Moss syndrome can be diagnosed by a battery of tests that measures the functioning of the middle ear (conductive loss battery).