Oculocerebrocutaneous (OCC) syndrome (OMIM 164180), a rare genetic disorder, is apparent at birth (congenital). The disorder is characterized primarily by eye (ocular), brain (e.g., cerebral), and skin (cutaneous) malformations. For example, many affected infants have semisolid or fluid-filled swellings (cysts) within the cavities of the skull (orbits) that accommodate the eyeballs and associated structures. In most patients, the eye on the affected side or sides is also abnormally small (microphthalmos). Brain abnormalities associated with OCC syndrome may include enlargement of the ventricular system, multiple fluid-filled spaces within and malformations of the outer region of the cerebral hemispheres (cerebral cortex), absence of the band of nerve fibers that joins the brain's hemispheres (agenesis of the corpus callosum), and a typical malformation of mid- and hindbrain. Affected infants and children often have intellectual disability and episodes of uncontrolled electrical activity in the brain (seizures). In addition, OCC syndrome is characterized by underdevelopment or absence of skin in certain localized regions (focal dermal hypoplasia or aplasia) and most have protruding, flesh-colored or brownish outgrowths of skin (cutaneous tags) within certain facial areas, including around the eyelids, on the cheeks, or near the ears. In all individuals with OCC syndrome known so far, the disorder occurs sporadically (with no family history of similar disorders). OCC syndrome has been reported more frequently in males than in females; however, the prevalence is unknown. Since the disorder was originally described in 1981 (JW Delleman & JWE Oorthuys), approximately 40 patients have been reported in the medical literature.