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A Report on Kaufman Oculocerebrofacial Syndrome

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Brief Report

Kaufman oculocerebrofacial syndrome is a disorder characterized by eye problems, intellectual disability and a distinctive pattern of facial features. Most individuals with Kaufman oculocerebrofacial syndrome have an unusually small head size and some have structural abnormalities of the brain. Affected individuals have weak muscle tone and are delayed in developing motor skills such as walking. Intellectual disability is severe or profound. Most affected individuals never acquire the ability to speak. Eye abnormalities and their effect on vision vary among people with Kaufman oculocerebrofacial syndrome. Some people with this disorder have abnormally small or poorly developed eyes; microcornea, in which the clear front covering of the eye is small and abnormally curved; missing pieces of tissue in structures that form the eye or underdevelopment of the nerves that carry signals between the eyes and the brain. Eyes that do not look in the same direction, nearsightedness or farsightedness, or an inward turning of the lower eyelid can also occur.

Kaufman oculocerebrofacial syndrome is caused by mutations in the *UBE3B* gene. This gene provides instructions for making a protein that plays a role in the ubiquitin-proteasome system, which is the cell machinery that breaks down unwanted proteins. The specific proteins that the *UBE3B* protein helps break down are unknown, but research suggests that *UBE3B* functions in the nervous system, digestive tract, respiratory system, and other organs and tissues, from before birth into adulthood. The *UBE3B* gene mutations that cause Kaufman oculocerebrofacial syndrome are thought to result in an abnormal *UBE3B* protein that cannot function properly or that is unstable and is rapidly broken down. Loss of this protein's function likely prevents cells from eliminating certain unnecessary proteins, resulting in problems with development and function of the brain, eyes, and other parts of the body.

Kaufman Oculocerebrofacial Syndrome (KOS) is characterized by severe intellectual disability and distinctive craniofacial features. Most affected children have prenatal-onset microcephaly, failure to thrive, hypotonia, and short stature. Eye abnormalities are common and can include structural abnormalities, refractive errors, strabismus, and entropion. Less common findings can include: unilateral or bilateral conductive hearing loss or mixed conductive-sensorineural hearing loss of variable severity; congenital heart defects; breathing problems; feeding difficulties; urogenital abnormalities; or skeletal abnormalities. Kaufman oculocerebrofacial syndrome is a rare autosomal recessive disorder which represents a phenotype mainly involving craniofacial and neurodevelopmental manifestations due to *UBE3B* gene mutations. The vast majority of the affected individuals exhibit microcephaly, eye abnormalities, and typical facial gestalt including blepharophimosis, ptosis, telecanthus, upslanting palpebral fissures, dysplastic ears, and micrognathia.

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