

Congenital fibrosis of the extraocular muscles (CFEOM) includes at least five rare genetic eye movement disorders present at birth that are characterized by incomitant strabismus. Specifically, there is an inability to move the eyes in certain directions (ophthalmoplegia), droopy eyelids (ptosis) and eyes that are fixed in an abnormal position. The oculomotor nucleus and nerve (cranial nerve III) and the muscles it serves and, in some cases the trochlear nucleus and nerve (cranial nerve IV) and/or the abducens nucleus and nerve (cranial nerve VI) and the muscles they serve are affected. Affected individuals have limited ability to move their eyes vertically (upward and downward) and can have variable limitations in moving their eyes horizontally. CFEOM is also frequently associated with droopy eyelids (ptosis) and eyes that are fixed in an abnormal position. Individuals with CFEOM often have their eyes fixed in a downward position, and elevate the chin so they can see. These disorders have been classified as CFEOM1, CFEOM2, and CFEOM3 based on ophthalmologic findings and molecular genetic testing. CFEOM3 can be characterized by additional involvement of the peripheral and central nervous system in addition to the eye findings. Tukel syndrome is characterized by missing and webbed fingers and toes in addition to the eye findings. These disorders do not worsen over time. CFEOM are rare disorders that have been seen in a range of diverse ethnic populations and affect males and females. A minimum prevalence has been estimated to be 1 in 230,000.