ISSN: 2573-9565

Review Article

Journal of Clinical Review & Case Reports

Congenital Fibrosis of the Extra-Ocular Muscles Syndrome

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Submitted: 01 Dec 2018; **Accepted**: 06 Dec 2018; **Published**: 15 Dec 2018

Abstract

CFEOM Syndrome is prevents the normal development and function of these muscles. As a result, people under the influence of the syndrome cannot move their eyes regularly. CFEOM1 syndrome and rare cases of CFEOM3 are caused by the mutation of the KIF21A gene, which is based on the long arm of chromosome 12, 12q12.

Keywords: CFEOM Syndrome, KIF21A Gene, Chromosome 12, Genetic Mutation

Generalizations of Congenital Musculoskeletal Syndrome (CFEOM) CFEOM syndrome is a genetic disorder that affects the muscles around the eye. These muscles control eye movements and eye position.

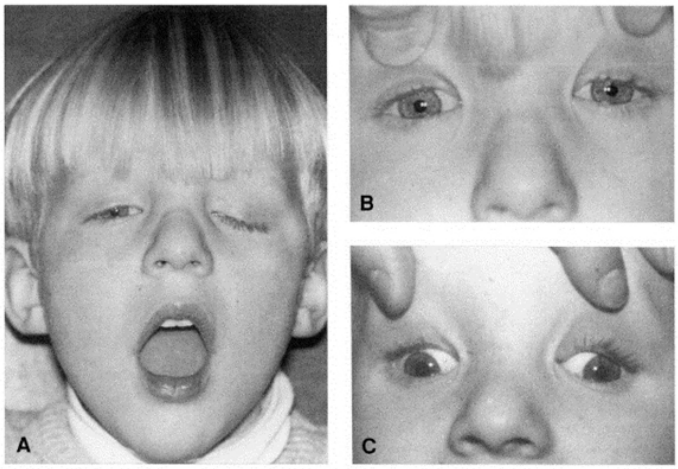


Figure 1: Picture of a child with CFEOM syndrome associated with related disorders in the eye.

Clinical Signs of Congenital Musculoskeletal Syndrome (CFEOM)

CFEOM syndrome prevents the normal development and function of these muscles. As a result, people under the influence of the syndrome cannot move their eyes regularly. Most people with this syndrome can look upwards with difficulty and may have limited eye movement. Additionally, people with CFEOM syndrome may have stomach ulcers (strabismus). Instead of moving the eyes to see moving objects, people with this syndrome may need to move their head to track those objects. In addition, many people with CFEOM syndrome have eyelid droopiness (petoisis), which often limits their vision.

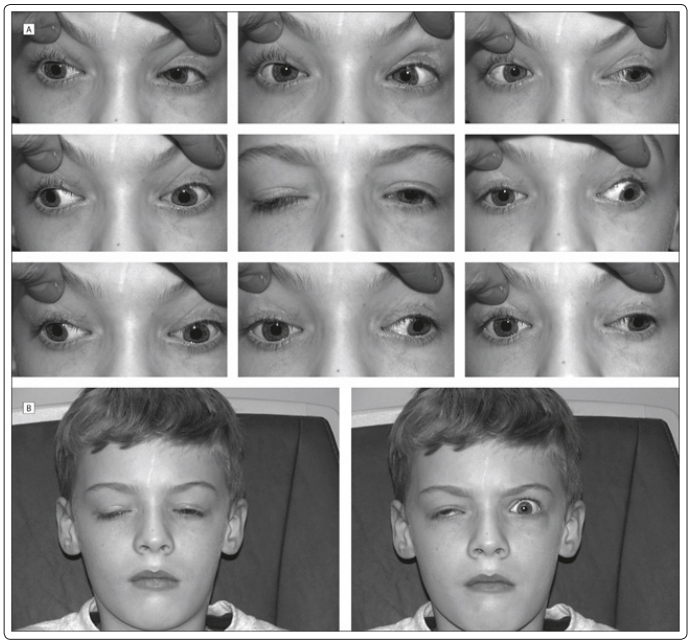


Figure 2: Another picture of eye disorders in patients with CFEOM syndrome.

Researchers have identified at least four forms of congenital fibrosis syndrome around the eyes that are known as CFEOM1, CFEOM2, CFEOM3, and Tukel syndrome. There are certain problems with eye movement between different types. Tukel syndrome is characterized by the loss of fingers (oligodactyly) and other hand abnormalities, in addition to eye movement problems.

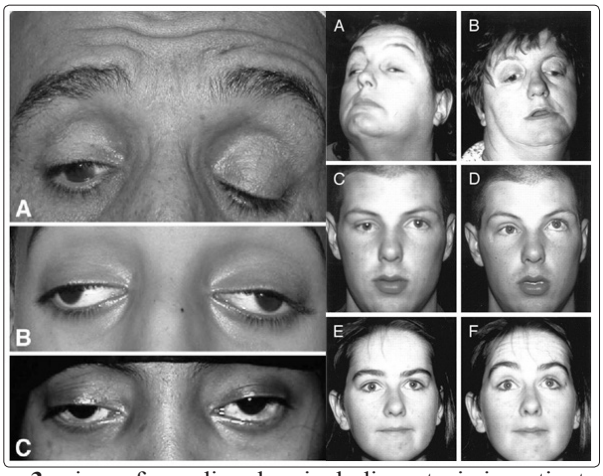


Figure 3: view of eye disorders including ptosis in patients with CFEOM syndrome.

Etiology of Congenital Musculoskeletal Syndrome (CFEOM)

CFEOM1 syndrome and rare cases of CFEOM3 are caused by the mutation of the KIF21A gene, which is based on the long arm of chromosome 12, 12q12. The gene provides instructions for the synthesis of a protein called quinine, which is essential for the transfer of materials in cells. Researchers believe that this protein plays an important role in normal growth and nerve function in the face and face. In particular, this protein plays an important role in the development of a specific branch of the third skull, which appears in the brain and controls the muscles of the eyes and the eyelids. The mutation in the KIF21A gene may change the ability of the quinine protein to convey material within the nerve cells, preventing the natural progression of these skull nerves and muscle control around the eyes. The abnormal function of the muscles around the eyes leads to limitation of eye movement and vision problems.

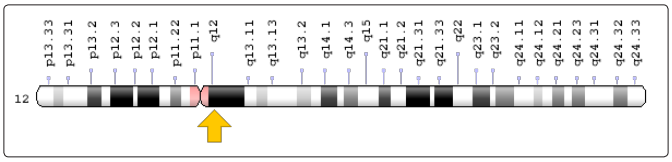


Figure 4: Schematic view of chromosome 12, which KIF21A gene is based on in the long arm of this chromosome 12q12.

The mutation in the PHOX2A gene, which is based on 11q13.4 in the long arm of chromosome 11, generates CFEOM2. This gene provides instructions for protein synthesis that can be found in the growing nerve system. Studies have shown that PHOX2A protein plays an important role in the development of third and fourth skeletal nerves that are essential for the normal movement of the eye. Mutations are likely to eliminate the function of the PHOX2A protein, which prevents the normal development of these skull and muscle nerves around the eye.

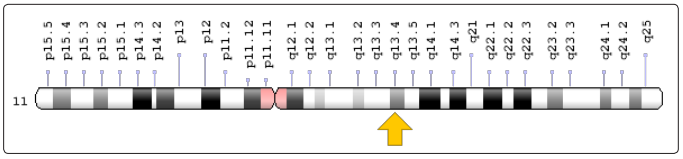


Figure 5: Schematic view of chromosome number 11, in which the PHOX2A gene is based on the long arm of this chromosome as 11q13.4

In most cases, CFEOM3 is the unknown cause of the disease. Studies have shown that a gene associated with CFEOM3 may be located near the end of chromosome 16. The gene associated with Tukel syndrome is also unknown, although the researchers think that it may be located near the end of chromosome 21.

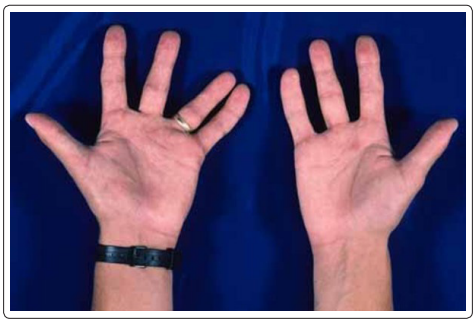


Figure 6: A picture of hands with CFEOM syndrome with oligodactyly and fingers curves

Congenital fibrosis syndrome follows the muscles around the eyes from different hereditary patterns. CFEOM1 and CFEOM3 follow the dominant autosomal inheritance pattern. Therefore, to produce these syndromes, a copy of the mutated gene KIF21A and the gene (?) (Parent or parent) are needed and the chance of having a child with this syndrome in the dominant autosomal state, for each possible pregnancy, is 50%.

Tukel syndrome and CFEOM2 syndrome follow an autosomal recessive pattern of heredity. Therefore, in order to produce these syndromes, two copies of the mutational gene of PHOX2A and the gene (?) (One of the father and the other of the mother) are needed and the chance of having a child with this syndrome in an autosomal recessive state, for any probable pregnancy 25%.

Frequency of Congenital Musculoskeletal Syndrome (CFEOM)

CFEOM1 is the most common form of congenital muscle fibrosis surrounding the eye, affecting at least 1 in 230,000 people. CFEOM1 and CFEOM3 have been reported worldwide, while CFEOM2 is seen only in a few families of Turks, Saudi Arabia and Iranian descent. Tukel syndrome appears to be extremely rare; it's known only to a large family of turkeys.

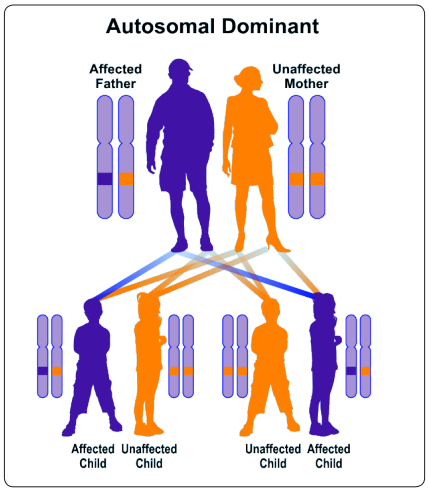


Figure 7: Schematic representation of the dominant autosomal inheritance pattern that follows the CFEOM1 and CFEOM3 syndrome.

Diagnosis of congenital muscle fibrosis syndrome around the eyes (CFEOM)

CFEOM syndrome is diagnosed based on the clinical and physical findings of the patients and some pathological and optological tests (medical optometry). The most accurate method for detecting this syndrome is the molecular genetic test for KIF21A, PHOX2A genes to investigate the presence of possible mutations.

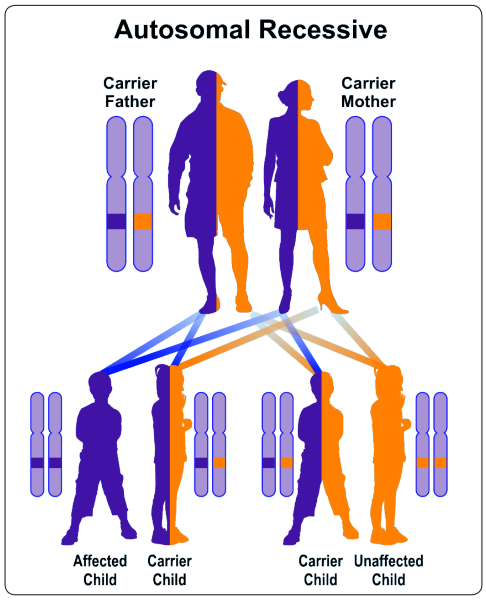
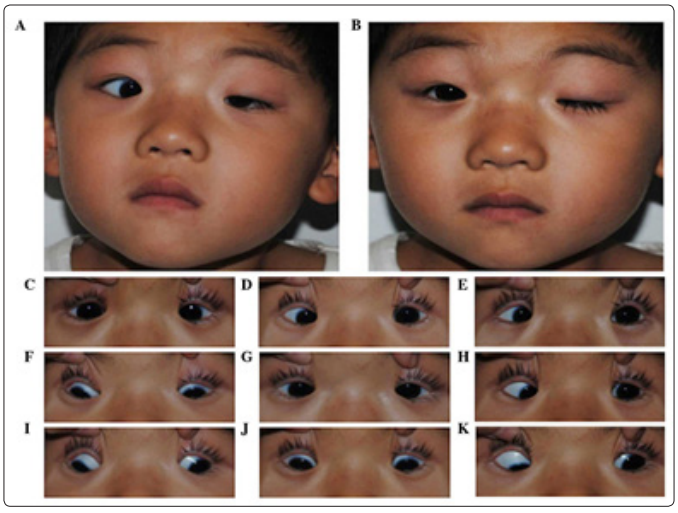


Figure 8: schematic view of an autosomal recessive hereditary pattern followed by CFEOM2 syndrome

Treatment pathways for the congenital muscle surrounding the eye (CFEOM)

The CFEOM syndrome treatment and management strategy is symptomatic and supportive. Treatment may be done by a team of experts, including eye specialists, neurologists, orthopedic surgeons, eye surgeons, and other health care professionals. There is no valid treatment for this syndrome, and all clinical measures are needed to reduce the suffering of the sufferers. Genetic counseling is also needed for all parents who want a healthy baby [1-13].



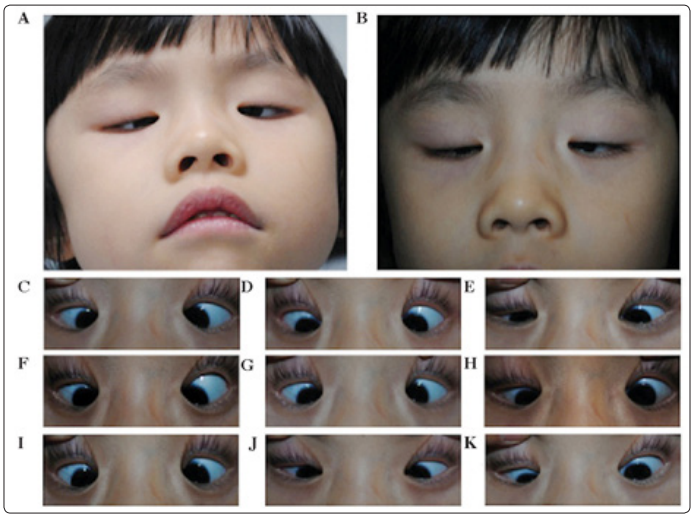


Figure 9: Overview of eye disorders in children with CFEOM syndrome.

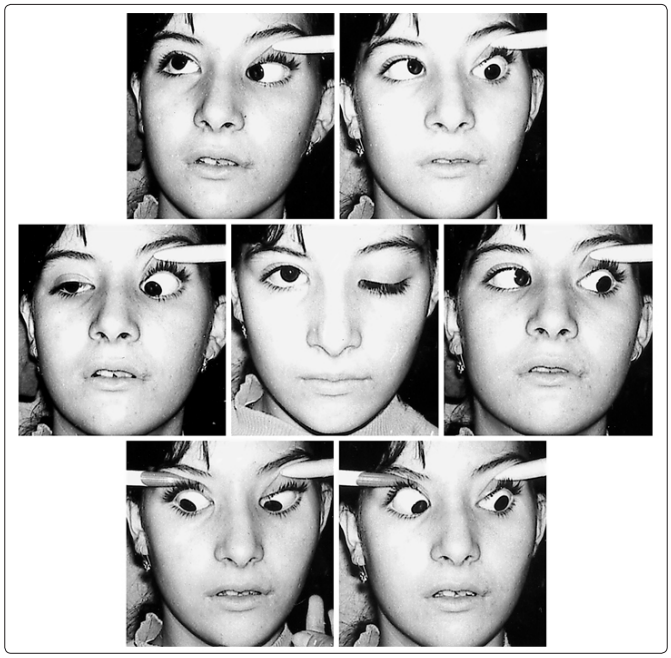


Figure 10: Images of ocular disorder in a person with CFEOM syndrome

Discussion and conclusion

This syndrome is one of the genetic disorders of the muscle that, unfortunately, there is no specific treatment for it. Patients suffering from the syndrome should endure the illness and their lifestyle with the disease until the end of their life. Scientists hope in the future to be able to use gene therapy to improve the condition of the patients with this syndrome

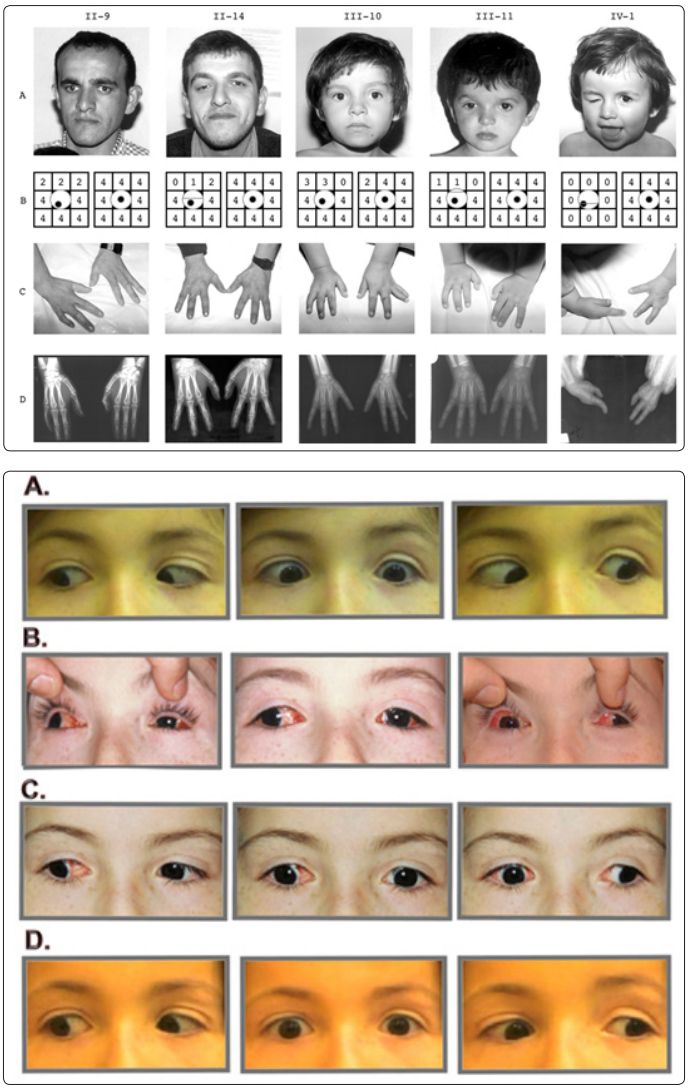


Figure 11: Images of relevant disorders in CFEOM syndrome

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