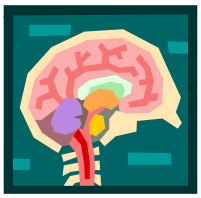

Disability Awareness Begins With You: Cerebro-Oculo-Facio-Skeletal Syndrome

What is Cerebro-Oculo-Facio-Skeletal Syndrome?

Cerebro-oculo-facio-skeletal syndrome (COFS) is a pediatric, genetic, degenerative disorder that involves the brain and the spinal cord. It is characterized by craniofacial and skeletal abnormalities, severely reduced muscle tone, and impairment of reflexes. Symptoms may include large, low-set ears, small eyes, microcephaly (abnormal smallness of the head), micrognathia (abnormal smallness of the jaws), clenched fists, wide-set nipples, vision impairments, involuntary eye movements, and mental retardation, which can be moderate or severe. Respiratory infections are frequent. COFS is diagnosed at birth. Ultrasound technology can detect fetuses with COFS at an early stage of pregnancy, as the fetus moves very little, and some of the abnormalities result, in part, from lack of movement.

NOTE: This disorder is not the same as Cohen's syndrome (cerebral obesity ocular skeletal syndrome).

Is there any treatment?

Treatment is supportive and symptomatic. Individuals with the disorder often require tube feeding. Because COFS is genetic, genetic counseling is available.

What is the prognosis?

COFS is a fatal disease. Most children do not live beyond five years.

What research is being done?

The NINDS supports research on genetic disorders such as COFS. The goals of this research include finding ways to prevent, treat, and cure these disorders.

Organizations:

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Information from: http://www.ninds.nih.gov/disorders/cofs/cofs.htm#What_is