Cerebrooculofacioskeletal Syndrome

National Cancer Institute

Source

National Cancer Institute. Cerebrooculofacioskeletal Syndrome. NCI Thesaurus. Code C3817.

A rare degenerative genetic disorder with an autosomal recessive pattern of inheritance that primarily affects the central nervous system. It is caused in some cases by mutations in the Cockayne syndrome group B gene, CSB/ERCC6, or the xeroderma pigmentosum genes: XPD/ERCC2, XPG/ERCC5, XPF/ERCC1, which are all involved in the transcription-coupled nucleotide excision repair pathway of DNA repair. It is currently thought to be part of the spectrum of disorders within Cockayne syndrome. Clinical signs at birth include microcephaly, hypotonia, abnormal reflexes and involuntary eye movements. The clinical prognosis is fatal with survivability beyond 5 years unlikely.