Cerebro-oculo-facio-skeletal (COFS) syndrome is a genetic degenerative disorder of the brain and spinal cord that begins before birth. The disorder is characterized by growth failure at birth and little or no neurological development, structural abnormalities of the eye and fixed bending of the spine and joints. Abnormalities of the skull, face, limbs and other parts of the body may also occur. COFS syndrome is inherited as an autosomal recessive genetic trait. COFS is now considered to be part of the spectrum of disorders within Cockayne syndrome. Cerebro-oculo-facio-skeletal syndrome is a very rare disorder present at birth. It affects males and females and occurs in many ethnic groups. The diagnosis of COFS syndrome is often made based on physical features seen at birth. X-ra. studies may reveal displacement of the small foot bones between the ankle and toes (second metatarsals) and neuroimaging studies may show reduced white matter with gray matter mottling. DNA repair studies on skin fibroblast cells may help to confirm a diagnosis of COFS syndrome. Molecular genetic testing to identify mutations in the excision repair genes is available on a research basis only.