Cockayne syndrome

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Identity

Inheritance: Autosomal recessive.

Clinics

Phenotype and clinics

Normal newborn; growth failure from the age of six months; diagnosis from the age of two years on:

Senile appearance of the skin (pigmentation, atrophy) with "mickey mouse" aspect (microcephaly, large ears, large nose, deep set eyes).

"Senil dwarf" aspect in contrast with long limbs, large hands and feet, cold fingers with cyanosis, flexion contractures of joints.

Sensitivity to sunlight.

Severe encephalopathy with profound mental retardation and sensory disorders (deafness, optic atrophy).

Pigmentary retinitis leading to cecity.

Other disorders: hypertension, early atherosclerosis, intracranial calcification, glomerulosclerosis.

Neoplastic risk

No increased susceptibility to skin tumors and other cancers, except for Cockayne syndrome expressing xeroderma pigmentosum (XP) symptoms (association with XPG, XPD or XPB group).

Evolution

Clinical heterogeneity, but early death from cachexia and dementia, early cutaneous tumors and atherosclerosis.

Cytogenetics

Inborn conditions

As in XP, the UV light-induced level of sister chromatid exchange (SCE) is increased as well as the rate of chromosome aberrations, mainly chromatid breaks.

Genes involved and proteins

Note

There is genetic heterogeneity in CS, giving rise to complementation groups.

The genes involved are: CSA, also called ERCC8 (ERCC for Excision-Repair Cross Complementing rodent repair deficiency) located on chromosome 5, CSB, also called ERCC6, located in 10q11-21; outside CSA and CSB, there is: 3 patients who are XPB/CS, involving XPB, also called ERCC3, located in 2q21; 2 patients XPD/CS, involving XPD, also called ERCC2, located in 19q13; and 6 patients XPG/CS, involving XPG, also called ERCC5, located in 13q32 (note: the class of patients with both XP and CS were classified earlier as CS III, but not anymore).

References


This article should be referenced as such:


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