Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Cancer Prone Disease Section

Mini Review

Cockayne syndrome

Claude Viguié

Service de Dermatologie, Hôpital Tarnier-Cochin, 89 rue d'Assas, 75006 Paris, France (CS)

Published in Atlas Database: October 2000

Online updated version : http://AtlasGeneticsOncology.org/Kprones/CockayneID10015.html
DOI: 10.4267/2042/37679

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 encephalopathia with profond 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 ligth-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

Brosh RM Jr, Balajee AS, Selzer RR, Sunesen M, Proietti De Santis L, Bohr VA. The ATPase domain but not the acidic region of Cockayne syndrome group B gene product is essential for DNA repair. Mol Biol Cell. 1999 Nov;10(11):3583-94

Bartenjev I, Butina MR, Potocnik M. Rare case of Cockayne syndrome with xeroderma pigmentosum. Acta Derm Venereol. 2000 May;80(3):213-4

de Boer J, Hoeijmakers JH. Nucleotide excision repair and human syndromes. Carcinogenesis. 2000 Mar;21(3):453-60

Hanawalt PC. DNA repair. The bases for Cockayne syndrome. Nature. 2000 May 25;405(6785):415-6

Rockx DA, Mason R, van Hoffen A, Barton MC, Citterio E, Bregman DB, van Zeeland AA, Vrieling H, Mullenders LH. UV-induced inhibition of transcription involves repression of transcription initiation and phosphorylation of RNA polymerase II. Proc Natl Acad Sci U S A. 2000 Sep 12;97(19):10503-8

This article should be referenced as such:

Viguié C. Cockayne syndrome. Atlas Genet Cytogenet Oncol Haematol. 2000; 4(4):222.