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COCKAYNE SYNDROME

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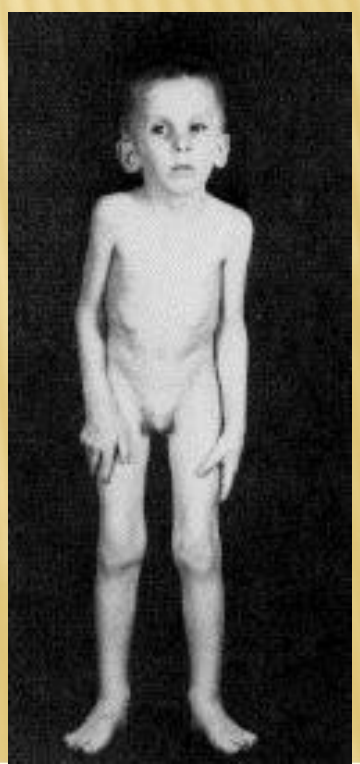
- Three types: I, II, III
- Appearance
  - Photosensitivity
  - Failure to gain weight/grow at expected rate
  - Microcephaly (small head)
  - Impaired development of the nervous system
  - Mental Retardation
  - Bone and eye abnormalities
- Multisystemic Degeneration
  - Premature aging
  - Hearing loss
  - Tooth decay
  - Changes in the brain



CLASSICAL DIAGNOSIS

- Type I (Classic C.S.)
  - Postnatal growth failure (<5th percentile by age 2)
  - Progressive neurologic dysfunction
  - Any of the aforementioned symptoms also taken into account
- Type II (Connotal C.S.)
  - Much more severe
  - Very little neurological development after birth
- Type III
  - Just recently confirmed as a form of C.S.
  - Much more mild

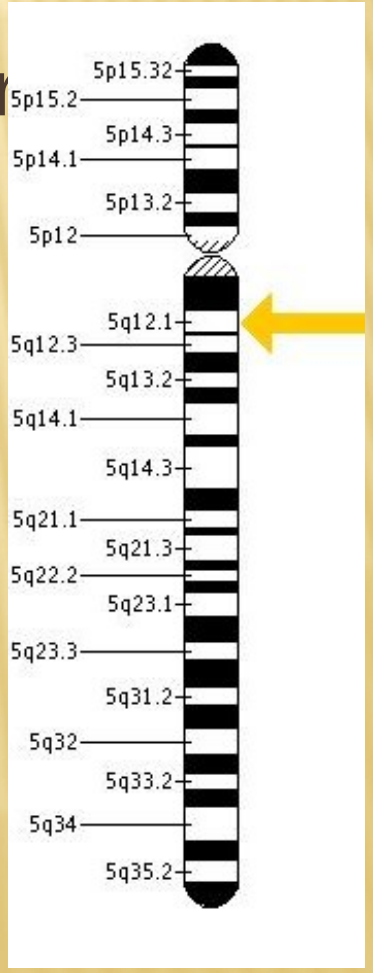
TYPE I COMPARED TO TYPE 2

Figure 1 - Patient A.P. at the age of 8 years, presenting typical features of Cockayne syndrome (thin face, slender nose, deep-set eyes and large ears).

- http://www.youtube.com/watch?v=RTIRucvW

MOLECULAR GENETICS

- Autosomal recessive
- Mutation of the ERCC 8 gene (25%) or ERCC 6 gene (75%)
- Located at 5q12.1 and 10q11.23, respectively
- Penetrance 100% in mouse models

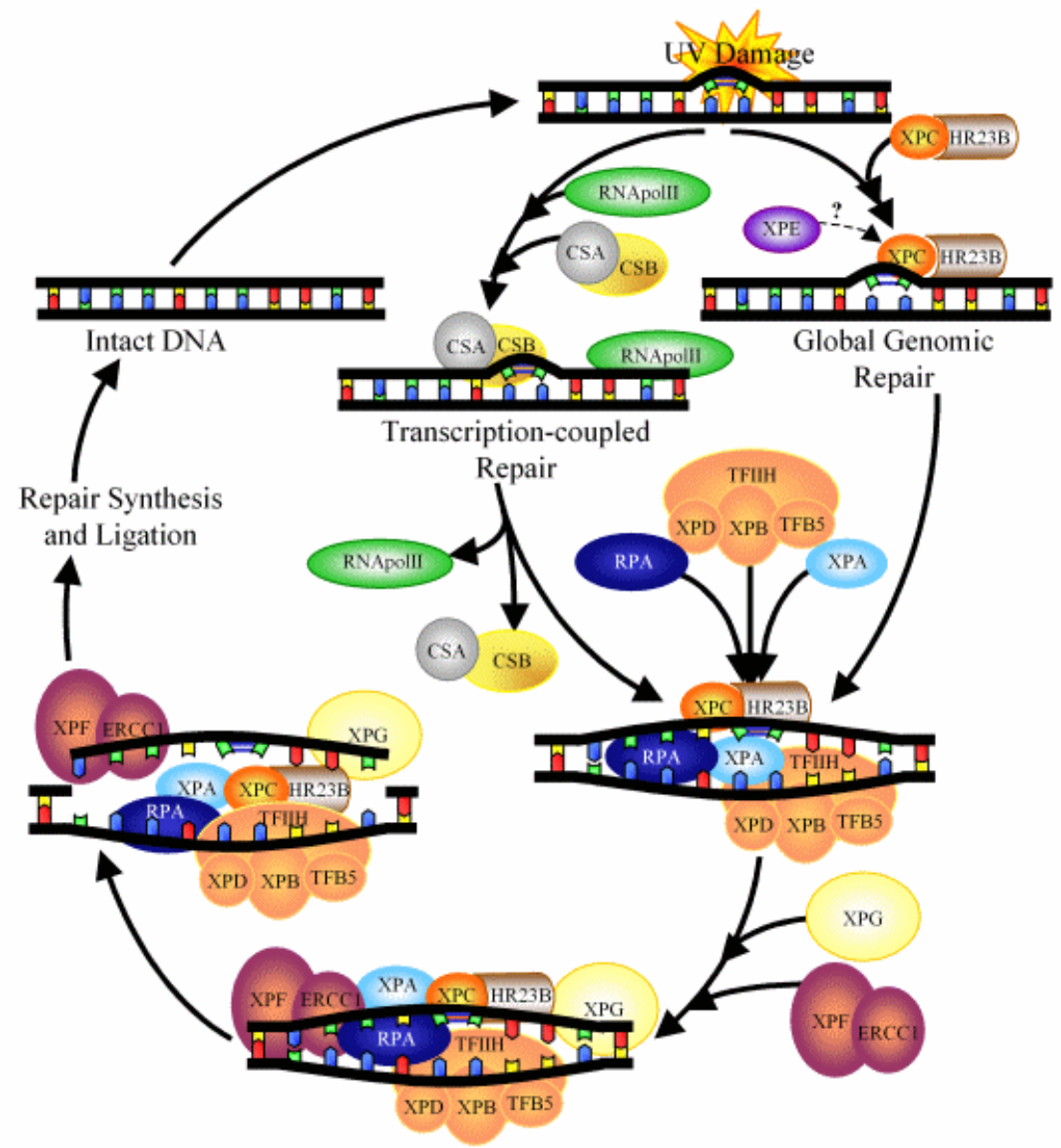


TREATMENT

- No treatment for the disorder
- Some light management of the symptoms, but nothing very effective
  - Physical Therapy
  - Feeding tube
  - Management of hearing loss and cataracts
  - Sunscreen

ERCC 6 AND 8 GENES

- Code for proteins that are involved in repairing damaged DNA



MODERN DIAGNOSIS

- DNA repair testing
- Confirmatory genetic testing
- Carrier testing
- Complementation tests to determine which gene is affected (for research basis only)
- Better therapies have yet to be developed.

SOURCES

- http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?boo
- http://mcb.asm.org/cgi/content/full/29/5/1276
- http://www.ncbi.nlm.nih.gov/sites/entrez?db=gene
- http://ghr.nlm.nih.gov/condition/cockayne-syndrom
- http://www.ncbi.nlm.nih.gov/omim/133540