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COCKAYNE SYNDROME
Cockayne Syndrome

- 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 (<5\textsuperscript{th} 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 1 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=RTIRucvV
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
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://mcb.asm.org/cgi/content/full/29/5/1276