Trichothiodystrophy (TTD)
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Identity
Alias: Ichtyosis, brittle hair, intellectual impairment, decreased fertility, and short stature syndrome (IBIDS)
Inheritance: Recessive autosomal.

Clinics
Phenotype and clinics
Photosensitivity, Ichtiosys, Brittle hair, Intellectual impairment, Decreased fertility, Short stature (PIBIDS syndrome).
Photosensitivity is absent in 50% of cases (therefore called IBIDS syndrome).

Neoplastic risk
This familial disease IS NOT a cancer prone disease but it involves the same complementation groups as in xeroderma pigmentosum and Cockayne syndrome (XPD, XPB), and share defects in similar genes.

Prognosis
Depends on the DNA repair defect (photosensitivity: XPD-ERCC2, XPB-ERCC3, TTD-A) and on the transcription errors (other signs).

Cytogenetics
Inborn conditions
No known chromosome abnormalities.

Genes involved and proteins
Note
The DNA repair defect is found in 3 classes:
Patient with TTD-A group (low level of the TFIIH transcription factor),
Patients mutated in the XPB gene (TTD/XPB), involving XPB, also called ERCC3, located in 2q21; and
All the other patients mutated in the XPD gene (TTD/XPD), involving XPD, also called ERCC2, located in 19q13.

References


This article should be referenced as such: