

Trichothiodystrophy



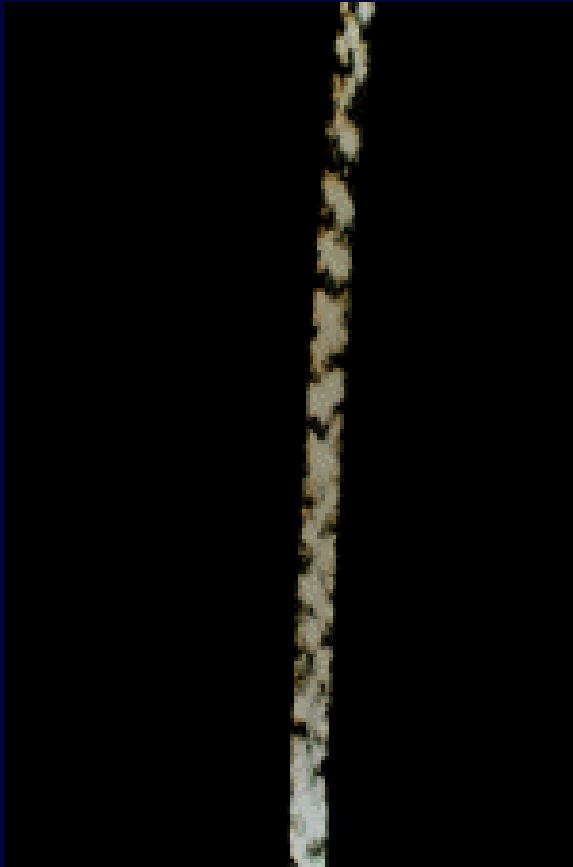
- Very rare
- First described in 1971 (Tay) in three Chinese siblings with intellectual disability, growth retardation and brittle hair
- Described in all ethnic groups

Trichothiodystrophy



- In 1980, brittle hair shown to be due to sulphur deficiency
- Name “trichothiodystrophy” first used

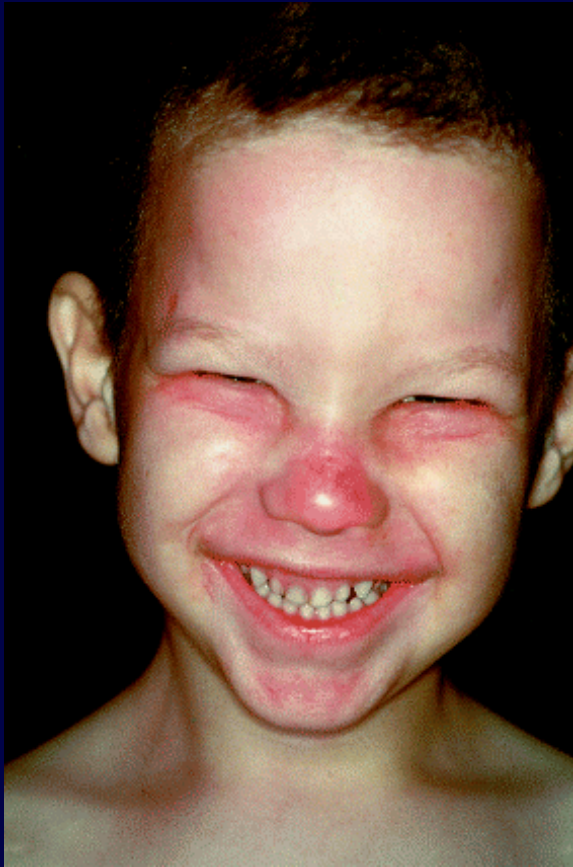
Trichothiodystrophy



TTD hair has characteristic “tiger tail” appearance

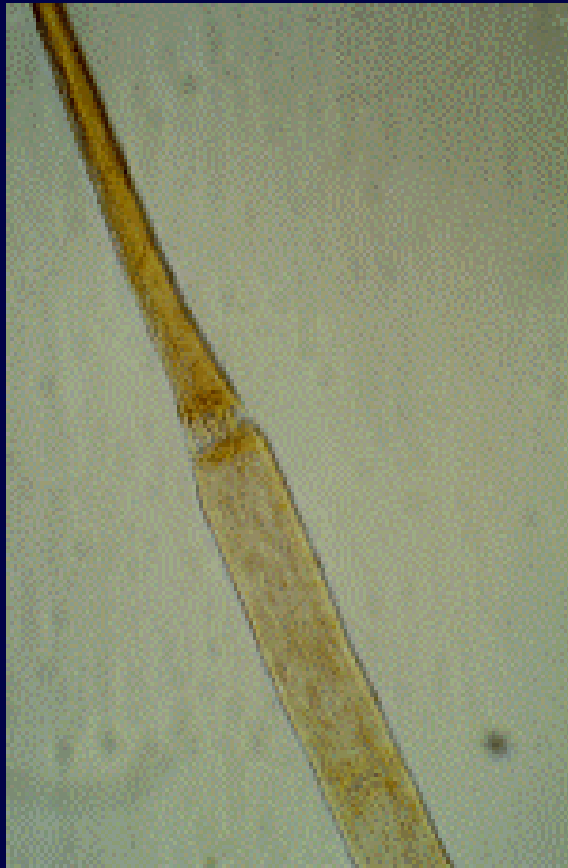
- TTD is now recognised as a group of related disorders that have in common ‘sulfur-deficient brittle hair’
- i.e. is TTD is more than one disorder

TTD Clinical Features I



- **Hair**
 - Brittle hair
- **Nails**
 - Brittle nails
- **Skin**
 - Scaly skin
 - Photosensitivity
 - Lack of subcutaneous fatty tissue
- **Growth**
 - Physical retardation
 - Low birth weight
 - Short stature
- **Neuro**
 - Mental retardation
- **Immunology**
 - Hypogammaglobulinemia
 - Recurrent infections

TTD Clinical Features II



- **Gastrointestinal**
 - Intestinal obstruction
- **Thorax**
 - Absent breast tissue
- **Genitourinary**
 - Hypogonadism
- **Face**
 - Aged appearance
- **Eyes**
 - Cataract
- **Head**
 - Microcephaly
- **Respiratory**
 - Asthma
- **Joints**
 - Joint contractures

TTD Management



- No specific treatment
- Photosensitivity may be extreme, even behind glass
- No increased incidence of skin cancer or internal malignancy
- Uncertainty about long term prognosis

TTD is inherited in an autosomal recessive manner

