

# CLOUSTON SYNDROME



## SYNONYM

Ectodermal dysplasia, hidrotic

**Clouston Syndrome is a Rare Genetic Condition Characterized by Abnormal Nails and Sparse Hair**

### Nails

- Severe malformation
- Spontaneous separation of the nail plate starting at the distal free margin and progressing proximally.
- Underdeveloped
- Thick, discolored

### Hair

- Fine, brittle, slow-growing
- Total balding (alopecia)
- Missing in spots for males
- Sparse eyelashes

- Sparse eyebrow
- Absent underarm hair
- Reduced hair strength

## **Additional Features Include:**

### **Skin**

- Normal sweating
- Abnormal thickening of the skin of the palms and soles (palmoplantar hyperkeratosis) that gets worse with age
- Hyperpigmentation on the knuckles, elbows, axillae, areolae, pubic area

### **Hands**

- Fluctuation and softening of the nail bed
- Increased convexity of the nail fold
- Thickening of the whole end part of the finger (distal) resembling a drumstick
- Shiny aspect and small grooves of the nails and skin

### **Eyes**

- Cataracts
- Light sensitivity (photophobia)
- Misalignment of the eyes (strabismus)
- Inflammation of the thin clear tissue that lies over the white part of the eye and lines the inside of the eyelid (conjunctivitis)
- Inflammation of the eyelids (blepharitis)
- Sparse eyelashes

- Sparse eyebrows

## **Growth**

- Short stature (usually mild)

# **DIAGNOSING CLOUSTON SYNDROME**

A physician can diagnose Clouston syndrome on the basis of physical features. Symptoms to look for in individuals include abnormal nails and sparse hair.

## **Cause of Clouston Syndrome**

Clouston syndrome is caused by mutations in the in the gap junction protein, beta-6 gene (GJB6).

## **Genetic Testing**

Genetic testing (<https://nfed.org/learn/diagnosis/genetic-testing/>) is available for Clouston syndrome.

## **Inheritance Pattern**

Clouston syndrome is inherited as an autosomal dominant disorder. An affected individual has a 50% chance of passing the condition on to each child.

Learn more about Clouston syndrome on OMIM - (Online Mendelian Inheritance in Man) (<https://www.omim.org/entry/129500?search=clouston&highlight=clouston>) or the Genetics Home Reference (<https://ghr.nlm.nih.gov/condition/clouston-syndrome>).

## **Additional Resources**

Nail Abnormalities in Clouston Syndrome (<https://www.nfed.org/learn/library/nail-abnormalities-clouston-syndrome/>)

## **Family Perspectives**

Clouston Syndrome: Perfectly Different (<https://nfed.org/blog/clouston-syndrome-perfectly-different/>)

How Clouston Syndrome Has Impacted Three Women's Fingernails and Life  
(<https://www.nfed.org/blog/clouston-syndrome-fingernails/>)

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