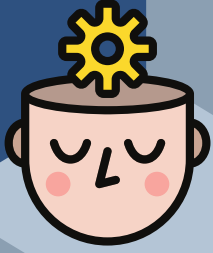


# CHARACTERISTICS OF A HIST1H1E INDIVIDUAL



## Hypotonia

- Low Muscle Tone
- Difficulty Feeding
- Delayed Milestones (Mild to Profound)
- Speech Disturbances (Delayed Speech, Apraxia, Non-Verbal)
- Strabismus (Crossed or Wandering Eyes)



## Intellectual Disability/Neurological

- Ranging from Mild to Profound
- Behavior Issues
  - ADHD
  - Autism Spectrum Disorder
  - Anxiety
  - Aggression (Not Common)
  - Sleep Disturbances (Early Rising)
- Seizures

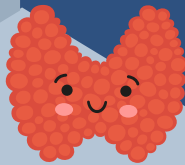
I



## Skeletal

- Scoliosis (Curvature of the Spine)
- Lower Limb Asymmetry
- Shortening of Fingers or Toes
- Permanently Bent Fingers or Toes
- Overlapping Toes
- Multiple Fractures (Rare)
- Premature Aging

S



## Testicular and Thyroid

- Hypothyroidism
- Cryptorchidism (Undescended Testicles in Males)
- Delayed Puberty

T



## Facial Features

- Early Childhood
  - Full Cheeks
  - High Hairline
  - Bitemporal Narrowing (Hollowing in Temples)
  - Deep-Set Eyes
  - Downslanting Palpebral Fissures (Eyes Appear Sad)
  - Wide Bridged Nose/Wide Set Eyes
  - Uprturned Broad Nose
  - Slightly Low-Set Ears
  - Large Head
- In Later Childhood and Adulthood
  - Notable High Frontal Hairline
  - Frontal Bossing (Prominent Forehead)

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## Cardiac Abnormalities

- Atrial Septial Defect

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## Abnormal Dentition

- Crumbling Teeth
- Tooth Decay
- Enamel Hypoplasia (Poor Enamel)
- Uneven Teeth

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## Ectodermal Problems

- Thin and/or Brittle Hair
- Slow Growing Hair
- Reduced Body Hair
- Thin/Brittle Nails
- Skin Nevi (Moles)

E

\*\*Please keep in mind that there is a very broad spectrum within the HIST1H1E individuals to this date. This list is meant to be informative of what parents and physicians have observed; it does not imply that every HIST1H1E individual will display all of these characteristics. We will continue to update as more information becomes available.\*\*



HIST1H1E  
Rahman Syndrome