HADD Syndrome

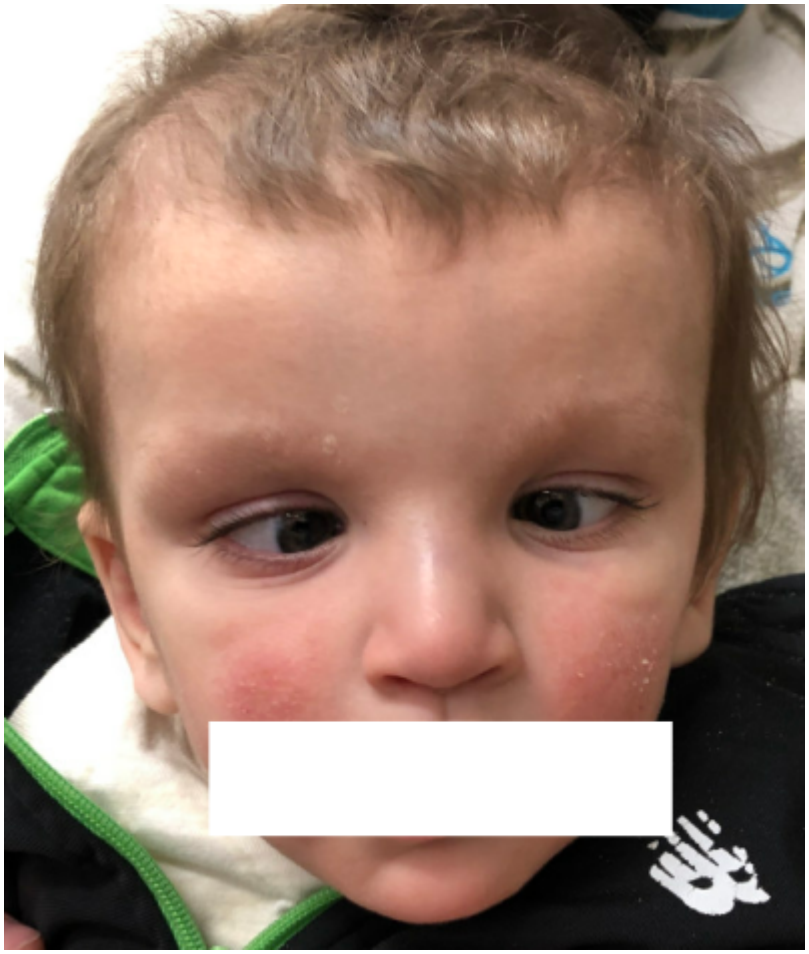
Hypotonia **A**taxia, **D**elayed **D**evelopment Syndrome AKA: EBF3 neurodevelopmental disorder (EBF3-NDD)

Main Features

- Intellectual disability
- Microcephaly
- Speech delay
    - mainly expressive speech delay
    - dysarthria
- Gait or truncal ataxia
- Hypotonia
- Behavioral problems
    - stereotypic movements (e.g., rotating movements, chewing on clothes, head retropulsion)
    - perseverative social behavior
    - short attention span
- Facial dysmorphism

Eye Findings

- Strabismus: Esotropia



- Deep-set eyes
- Synophrys, Straight eyebrows
- Astigmatism
- Epicanthal folds
- Hypertelorism
- Downslanting palpebral fissures

Other Findings

- Genitourinary
  - micropenis
  - cryptorchidism
  - vesicoureteral reflux
  - renal anomalies
- Gastrointestinal abnormalities
- Musculoskeletal abnormalities
- MRI Brain findings
  - Cerebellar vermis hypoplasia
  - Cerebellar atrophy or hypoplasia
  - Small inferior posterior cerebellar lobes and hypoplasia of the posterior vermis with mild prominence of the ventricles and sulci
  - Abnormal configuration of cerebellar folia arranged in radial shape

Etiology

- EBF3 gene mutation

Reference

- Gene Reviews
- Med Gen Summary
- OMIM

syndrome