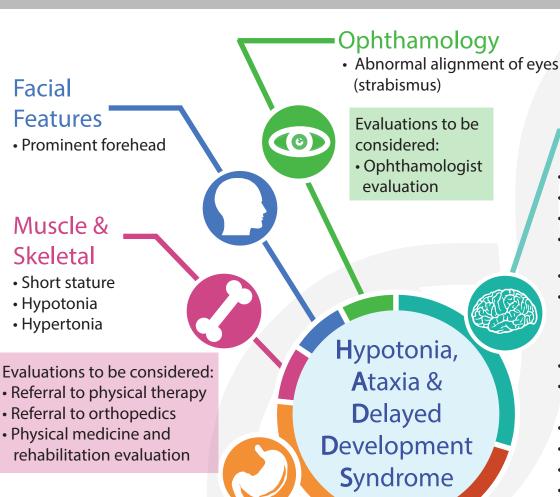
What is EBF3-Related HADDS?

NOTE:

This infographic serves to illustrate common features in *EBF3* HADDS patients as observed through the study and as a tool to guide discussions with caregivers and healthcare providers.



Gastrointestinal

- Constipation
- Gastroesophageal reflux
- Feeding difficulties

Evaluations to be considered:

- Swallow study
- Motility study
- Gastroenterologist evaluation

Additional evaluations may be considered as clinically indicated.

Genitourinary

- Recurrent urinary tract infection (UTI)
- Bladder control problems

Evaluations to be considered:

- Renal/Bladder imaging
- Basic labs (CBC, chemistries)
- Voiding study
- Urologist evaluation

Neurology

- Low muscle tone
- Motor delay
- Motor incoordination
- Speech delay
 - Speech apraxia
- Hypomimia
- Autistic features
 - · Aversion to crowds and loud noise
 - Poor eye contact
 - Motor stereotypies
- Seizures
- Ataxia
 - Unstable or wide-based gait
- Brain MRI anomalies
- Weak or absent cry in infancy
- Repetitive itching
- Decreased sensitivity to pain

Evaluations to be considered:

- Brain MRI scan
- Neuropsychology evaluation
- Physical, speech, and occupational therapy
- Neurologist evaluation

Not all features may present themselves in patients.