Neural Degenerative Disorders: Tay-Sachs Disease

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(March 16, 2016)

[Image: Source: Boston, 1995]

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Figure 3: known locations of mutations that disrupt HexA activity, red—acute to subacute, green—chronic, cyan—asymptomatic[1]

Timeline of Symptom Presentation

- Early Fetal Stages
  - Destruction and interruption of normal neural tissue development and activity begin
- 0-6 Months of Age
  - Near normal physical presentation
  - Clear onset of neurological damage
  - Slowed development
  - 0.5-2 Years of Age
    - Recurrent seizures
    - Substantially reduced mental functionality
    - Regression of infant (loss of ability to crawl, sit up, turn over)
  - 2-4 Years of Age
    - Blindness
    - Paralysis, limited or no response to external stimuli
  - 5 Years of Age
    - TSD is typically fatal at this point

Possible Treatments for TSD

- No curative treatment yet exists for TSD, so all care is palliative (treats symptoms to slow deterioration) (Mayo Clinic, 2016)
- Anti-epileptics (AEDs) for seizures
- Respiratory medication for breathing
- Funding for Genetic Therapeutics (Allison & Hall, 2005)
- CPT for development of new treatments
- Drugs, chemical therapeutics, and vector-based gene therapies cannot effectively reach diseased cells because of blood-brain barrier (Guyton & Hall, 2005)
- N-butyldeoxyxojirimycin to prevent somal storage of lipids (Platzek et al., 1997)
- Engraftment of transduced progenitor/stem cells (Lacerenza et al., 1996)
- Continuing development of genetic therapeutics to correct for mutated gene using retrovirus/adenoviral vectors, CRISPR-Cas9, Zinc Finger Nucleases (ZFNs), TALEN (Cosgrove, 2016)
- Assay for looking for increased expression of both functional and non-functional forms in mouse cells (Gruber et al., 1999)
References


