The Long and Winding Road: A Case Study of a Child with Niemann-Pick Disease

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The Long and Winding Road: A Case Study of a Child with Niemann-Pick Disease

Lehigh Valley Reilly Children’s Hospital: Children’s Cancer and Multipurpose Infusion Center

Lehigh Valley Health Network, Allentown, PA

OBJECTIVES
1. Describe the clinical characteristics and key care considerations for the child with Niemann-Pick Disease (NPD).
2. Share three pragmatic tactics that can be implemented to promote quality of life in pediatric patients affected by Niemann-Pick Disease.

CASE PRESENTATION
- 8 year-old Caucasian female
- Symptoms began at age 3
- Falls with laughter (Cataplexy)
- Hepatomegaly
- Splenomegaly
- Abnormal eye movements
- Hearing loss
- Behavioral manifestations
- Diagnosed at age 4

UNCOVERING THE DIAGNOSIS
- Laboratory Testing – CBC, reticulocyte count, HDL, LDL, Triglycerides, lipid profile – normal
- CMP – slightly elevated AST

GENETIC TESTING:
Patient – Identified one deleterious mutation and one variant of uncertain significance. Genetic testing does not confirm NPD – skin biopsy recommended for disease diagnosis.
Parents – Niemann Pick Type C (NPC) gene carriers

INITIAL AND SECONDARY SKIN BIOPSIES – CHILD:
Positive filipin staining with reflex cholesterol esterification – confirmed the NPC diagnosis

DETECTION
- Diagnosis is often delayed – may impede initiation of crucial therapies
- Parents
- Bone marrow
- Liver
- Lungs
- Brain
- Spleen
- Bone marrow
- Low disease awareness
- Varied onset
- Classic presentation mid to late childhood
- Heterogeneous visceral, neurological and psychiatric symptoms:
  - Seizures
  - Hearing loss
  - Jaudice
  - Developmental delay
  - Declining academic performance
- 4 subclasses – Type C is most common
- Splenomegaly
- Dystonia
- Ataxia
- Vertical gaze abnormalities
- Dementia
- Incidence – 1:120,000 live births
- Diagnosis is often misdiagnosed, underdiagnosed or delayed – may impede initiation of crucial therapies

TREATMENT/MANAGEMENT
TREATMENT PROTOCOL
- TWICE DAILY MIGLUSTAT 100mg oral capsule
  - May slow the progression of the disorder by limiting the production of glycolphingolipids – not curative
- BI-WEEKLY LUMBAR PUNCTURE WITH INTRATECHAL VITSESSO-270 (VTS-270)
- VTS-270:
  - Targets chemicals stored in cells such as cholesterol and sphingolipid
  - Minimizes accumulation of cholesterol in the brain that can cause damage and death of cells by bypassing the broken NPC1 pathway – restores normal metabolism and cell regulation

REFERENCES
http://www.uptodate.com/contents/overview-of-niemann-pick-disease

KEY TAKE-AWAYS
- Intrathecal administration is required as medication does not cross the blood brain barrier
- Medication side effects
  - Miglustat-Diarrhea, flatulence, abdominal pain
  - VITSESSO-270 High frequency hearing loss

INTRATECHAL TREATMENT SIDE EFFECTS
- Back pain, intermittent headaches due to bi-weekly lumbar punctures

SUPPORTIVE THERAPIES
- Physical and occupational therapy

INTERPROFESSIONAL REFERRALS
- Neurology, Audiology, POC, metabolic disease specialist, gastroenterologist, annual visits to the National Institute of Health (NIH) for NPD research study monitoring

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