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

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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

WHAT IS NIEMANN-PICK?

- Also called Childhood Alzheimer's
- Rare, fatal
- Progressive neurodegenerative disorder
- Autosomal recessive inheritance pattern
- Impairs lipid metabolism – damages cells in the:
  - Spleen
  - Liver
  - Lungs
  - Brain
  - Bone marrow
- Low disease awareness
- Varied onset
- Classic presentation mid to late childhood
- Heterogeneous visceral, neurological and psychiatric symptoms:
  - Seizures
  - Hearing loss
  - Jaundice
  - Developmental delay
  - Declining academic performance
  - Hepatomegaly
  - 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
- 4 subclasses – Type C is most common

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

TREATMENT/MANAGEMENT

TREATMENT PROTOCOL

- Twice daily Miglustat 100mg oral capsule
  - May slow the progression of the disorder by limiting the production of glycophingolipids – not curative
- Bi-weekly lumbar puncture with Intrathecal VTESSE-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
  - Intrathecal administration is required as medication does not cross the blood brain barrier

MEDICATION SIDE EFFECTS

- Miglustat-Diarrhea, flatulence, abdominal pain VTESSE-270 high frequency hearing loss

INTRATHECAL TREATMENT SIDE EFFECTS

- Back pain, intermittent headaches due to bi-weekly lumbar punctures

SUPPORTIVE THERAPIES

- Physical and occupational therapy

INTERPROFESSIONAL REFERRALS

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

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KEY TAKE-AWAYS

Nurses are in a unique position to assist the patient and family manage the physical, psychosocial and care partnership needs prompted by NPD:

PHYSICAL:

- Ensure the patient feels safe
- Encourage therapeutic play

PSYCHOSOCIAL:

- Provide consistent nursing care
- Maintain an open relationship with the family
- Educate the patient/family on current disease information

CARE PARTNERSHIP:

- Promote interprofessional collaboration to optimally manage a child diagnosed with NPD

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