Patient Name: Emily Johnson
DOB: 2019-03-14
MRN: 04273928
Date of Visit: 2025-07-29
Provider: Dr. Jane Smith, MD – Pediatric Neurology
Diagnosis: Phelan-McDermid Syndrome (22q13 deletion syndrome)

Chief Complaint:
Developmental delay, limited expressive language, behavioral concerns, and hypotonia.

History of Present Illness:
Emily is a 6-year-old girl with a confirmed diagnosis of Phelan-McDermid Syndrome. Her parents report global developmental delay, nonverbal communication, frequent hand-flapping, and reduced social interaction. Mild sleep disturbances and occasional feeding issues are present. No known history of seizures. She receives weekly speech, occupational, and physical therapy.

Medications:

- Melatonin 1mg at bedtime
- Multivitamin daily

Physical Exam Summary:

- Head circumference at 10th percentile
- Cardiac and respiratory exams normal
- Generalized hypotonia, mild gait instability
- Limited eye contact, minimal verbal output

Assessment:
Presentation is consistent with PMS. Emily displays hallmark features including hypotonia, developmental delays, and autistic-like behaviors. No acute medical concerns today.

Plan:

- Continue current therapies (SLT, OT, PT)
- Refer to developmental pediatrician and sleep specialist

- Monitor for emerging seizures or regression
- Follow up in 6 months or sooner if needed
- Offer genetic counseling

Parent Education:
Provided education on PMS, prognosis, and available resources. Directed family to the Phelan-McDermid Syndrome Foundation and shared printed materials.