Clinical Care Practice Parameters for Medical Assessment and Monitoring in Phelan-McDermid syndrome

What patients and doctors should talk about

This diagram identifies the areas of medical specialties that may be helpful in evaluating a person diagnosed with Phelan-McDermid syndrome (PMS), the condition(s) that may be present, and the tests that may help with diagnosis.

Psychiatry & Psychology
- Autism Spectrum Disorder
- Maladaptive behavior
- Intellectual Disability
- Absent or delayed speech
- Gold Standard diagnostic assessments
- Psychiatric Evaluation
- Cognitive and adaptive behavior testing

Neurology
- Seizures
- Structural brain abnormalities
- Feeding difficulties
- Hypotonia
- Motor skills deficits
- Decreased perception of pain
- Overnight video electroencephalography
- Brain imaging and head circumference monitoring
- Feeding therapy evaluation
- Occupational & physical therapy evaluations

Endocrinology
- Short/tall stature
- Hypothyroidism
- Temperature Regulation
- Monitor height, weight, and body mass index
- Metabolic work-up, including thyroid function
- Nutritional assessment

Nephrology
- Vesicoureteral reflux
- Urinary tract infections
- Hydonephrosis
- Renal cysts, hypoplasia, or agenesis
- Renal and bladder ultrasonography
- Voiding cystourethrogram
- Monitoring blood pressure

Cardiology
- Congenital heart defect
- Electrocardiography
- Echocardiography

Gastroenterology
- Gastroesophageal reflux
- Constipation/diarrhea
- Pica
- Referral for dietary changes and/or medication
- Bowel regimens
- Referral to behavioral therapy

Primary Care
- Upper respiratory tract infections
- Recurring ear infections
- Hearing and vision problems
- Lymphedema
- Decreased perspiration/heat intolerance
- Dental Hygiene
- Careful and consistent monitoring and management
- Referral to otolaryngology, ophthalmology, physiatry, dental and orthopedics

This document is a guide; NOT all patients exhibit every symptom or need all specialists or all tests.

Adapted from:
Kolevzon et al. (2014), Journal of Neurodevelopmental Disorders (www.jneurodevdisorders.com/content/6/1/39)