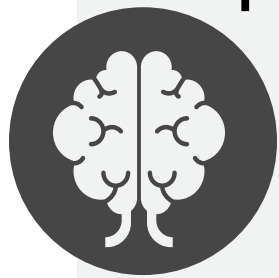


Clinical Care Practice Parameters for Medical Assessment and Monitoring in Phelan-McDermid Syndrome

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.

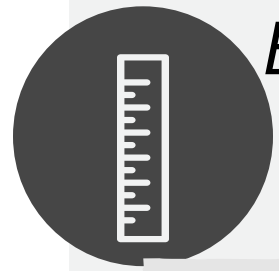
Psychiatry & Psychology



- Autism Spectrum Disorder
- Maladaptive behavior
- Intellectual Disability
- Absent or delayed speech

- Gold Standard diagnostic assessments
- Psychiatric Evaluation
- Cognitive and adaptive behavior testing

Endocrinology



- Short/tall stature
- Hypothyroidism
- Temperature Regulation

- Monitor height, weight, and body mass index
- Metabolic work-up, including thyroid function
- Nutritional assessment

Cardiology



- Congenital heart defect

- Electrocardiography
- Echocardiography

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, psychiatry, dental and orthopedics

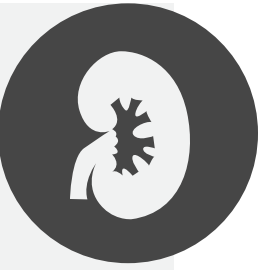
Neurology



- 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

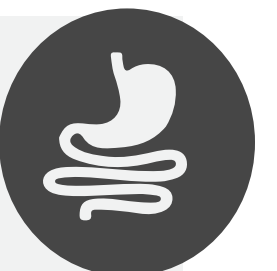
Nephrology



- Vesicoureteral reflux
- Urinary tract infections
- Hydronephrosis
- Renal cysts, hypoplasia, or agenesis

- Renal and bladder ultrasonography
- Voiding cystourethrogram
- Monitoring blood pressure

Gastroenterology



- Gastroesophageal reflux
- Constipation/diarrhea
- Pica

- Referral for dietary changes and/or medication
- Bowel regimens
- Referral to behavioral therapy

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



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v2.

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(www.jneurodevdisorders.com/content/6/1/39)

Phelan, et al. 2018. GeneReviews® Phelan-McDermid Syndrome
06/07/18 (<https://www.ncbi.nlm.nih.gov/books/NBK1198/>)