Phelan-McDermid Syndrome - Neuropsychiatric Consultation Group

Management of Symptoms

Case Presentation Form

Mount Sinai Seaver Autism Center for Research and Treatment

Complete this form and email to Dr. Alex Kolevzon at alexander.kolevzon@mssm.edu

Reset

Presenting Provider Name: _____

Clinic/Facility Name & City: _____

Provider Phone Number: _____

Provider Email Address: _____

ECHO ID: [ ]

Biological Gender: [ ]

Presentation Type: [ ]

Patient Age: [ ] (Yrs) [ ] (Mos)

Presentation date: _____

Race:
(Hold control and hold to select more than one race)

- American Indian/Alaskan Native,
- Asian,
- Black/African American,
- Native Hawaiian/Pacific Islander,

Other: _____

Ethnicity:

- Hispanic/Latino
- Not Hispanic/Latino
- Prefer not to say

When we receive your case, we will email you with a confidential patient ID number (ECHO ID) that must be utilized when identifying your patient during clinic. Please do not include any protected health information.

PLEASE NOTE: Project ECHO® case consultations do not create or otherwise establish a provider-patient relationship between any ECHO PMS-NCG clinician and any patient whose case is being presented in a Project ECHO® setting.

Project ECHO®

PMS - Neuropsychiatric Consultation Group

PLEASE NOTE that ECHO PMS-NCG case consultations do not create or otherwise establish a provider-patient relationship between any team clinician and any patient whose case is being presented in an ECHO PMS-NCG setting.

What problem(s) would you like help with for your patient?
(A longer narrative on the evolution of the problem(s) is welcome on a separate page or two attachment)

Does this child have a diagnosis of PMS?

If Yes, age at diagnosis:

Type of Deletion or Mutation (if known):

De novo status confirmed?

Does this child have an autism diagnosis?

If Yes, age at diagnosis:

ECHO ID:

PMS - Neuropsychiatric Consultation Group

Communication ability

- ☐ Non-verbal (no functional words)
- ☐ Uses single words
- ☐ Uses 2-3 word phrases
- ☐ Uses sentences
- ☐ Reciprocal conversation
- ☐ Follows 1-2 step directions
- ☐ Uses gestures

IQ (if available):

Behavior Concerns

- ☐ Anxious or worries
- ☐ Short attention span
- ☐ Hyperactivity
- ☐ Obsessive-compulsive symptoms
- ☐ Aggressive
- ☐ Unusual or excessive fears
- ☐ Depression
- ☐ Mood Lability
- ☐ Self-Injury (head banging, biting, scratching, cutting, picking, etc.)
- ☐ Toileting issues, accidents
- ☐ Catatonia (unusual postures, new motor patterns, etc.)
- ☐ Sleep disturbance (see sleep history on pg. 5)
- ☐ Possible hallucinations or other psychotic symptoms

Please tell us about the child's developmental and behavioral concerns. Please be as specific as possible:

ECHO ID:

Developmental History (cont.)

Has there been a period of **significant loss** of acquired skill(s)?

If so, did the child subsequently recover some or all of those skills?

Explain:

Medical/Psychiatric History: Please list all diagnoses or illnesses

How long has the child been in your care?

Diagnosis/Illness	Age

Current Medications

Medication	Dosage	Age when started	Reason for medication	Is it helping?

Please check all of the following that apply:

- ☐ Seizures
- ☐ Heart Problems
- ☐ Constipation
- ☐ Nausea/vomiting
- ☐ Diarrhea
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Gastroesophageal Reflux
- ☐ Kidney Problems
- ☐ Lung Problems
- ☐ Sleep disturbance
- ☐ Chronic Infections
- ☐ Environmental allergies
- ☐ Skin problems (rash, eczema)
- ☐ Gastroparesis, other GI dysmotility
- ☐ Autonomic instability affecting temperature, blood pressure or perfusion

ECHO ID:

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Laboratory Test	Performed	Results
Chromosomal Microarray		
Karyotype		
Sequencing Panel		
Whole Exome Sequencing		
MRI of the brain		
EEG		
Sleep Study		
Audiologic exam		
EKG/Echocardiogram		
Renal Ultrasound		
Metabolic work-up		
Immunologic work-up		

Dietary/Nutrition/Metabolic

Current height: _____ **Current weight:** _____

Please check all of the following that apply:

- ☐ Problem eater
- ☐ Picky eater
- ☐ Difficulty with solids
- ☐ Special Diet
- ☐ Eating/craving non-food items
- ☐ Difficulty with liquids
- ☐ G/J Tube in place

Types and amouts of liquids:

Sleep History

Rarely = never or 1 time/week; **Sometimes** = 2-4 times /week; **Usually** = 5 or more times/week

Does child...?:	How often?	Is it a problem?
Fall asleep within 20 minutes?		
Co-Sleep (With whom?)		
Awaken more than once during the night?		
Snore loudly		
Seem tired during the day		

Has the child had distinct periods during which sleep patterns have changed markedly? Explain:

Sleep comments:

ECHO ID:

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Trauma/Abuse History

Trauma /serious accidents _____ Physical Abuse _____ Sexual Abuse _____

Social History

Who is living in the home?

Relationship (1/2 sib, step parent, etc.)	Age

Biological parents are:

Siblings/Other Pregnancies:
(*Include any miscarries, stillbirths, or babies that died)*

Family History

Condition/Disorder	Mom	Dad	Brother	Sister	Mat GM	Mat GF	Pat GM	Pat GF
Autism Spectrum Disorder	☐	☐	☐	☐	☐	☐	☐	☐
Intellectual disability	☐	☐	☐	☐	☐	☐	☐	☐
Learning disability	☐	☐	☐	☐	☐	☐	☐	☐
Seizure disorder (epilepsy)	☐	☐	☐	☐	☐	☐	☐	☐
Psychiatric disorders	☐	☐	☐	☐	☐	☐	☐	☐
Childhood deaths	☐	☐	☐	☐	☐	☐	☐	☐
Genetic syndromes	☐	☐	☐	☐	☐	☐	☐	☐

ECHO ID: _____

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Educational History

What best describes the child's current education program?

- ☐ Full time in regular education class
- ☐ Full time in substantially separate special education class or school
- ☐ Time split between regular and special education classes
- ☐ Aide/Paraprofessional or extra help
- ☐ Home School
- ☐ Participation in Birth - 3 Early Intervention Programs (AR First Connections) Early
- ☐ Childhood Special Education - Local Educational Cooperative (3-5 yr)

Related Services

- ☐ Behavioral Therapy/ABA
- ☐ Physical Therapy (PT)
- ☐ Counseling
- ☐ Speech Language Therapy (SLT)
- ☐ Occupational Therapy (OT)
- ☐ Alternative Therapies

Other:

ECHO ID: