

## **PHELAN-McDERMID SYNDROME EMERGENCY CARD**

### **HEALTH CARE PROFESSIONALS INFORMATION ABOUT PHELAN-McDERMID SYNDROME (PMS)**

#### **General information**

Phelan McDermid syndrome (PMS) is a clinically variable disorder, mainly characterized by intellectual disability (mostly moderate to severe), absent or severely delayed speech, behaviour that may include autism characteristics, and a variety of other signs and symptoms. Typically, PMS is caused by a deletion of chromosome 22, including band 22q13.33, or a pathogenic variant in *SHANK3*.

Listed below are the features that are important in an emergency situation. For a full overview of all features see Schön et al., EJMG 2023.

#### **Frequently occurring problems (>30%)**

- Developmental delay/Intellectual disability
- Marked speech impairment
- Hypotonia
- Decreased pain response
- Hypohidrosis\*
- Autism spectrum disorder
- Hyperactivity<sup>#</sup>
- Sleeping problems<sup>#</sup>
- Regression
- Cyclical mood disorders
- Gastro-intestinal problems (constipation, diarrhoea)
- Dysmorphisms (a.o. long eyelashes, ptosis, broad nose, pointed chin, ear anomalies, malocclusion, retrognathia, large fleshy hands)

#### **Less frequently occurring problems (<30%)**

- Seizures
- Vision disturbances, including strabism
- Hearing loss
- Aggression against others and self
- Gastro-oesophageal reflux
- Cardiac anomalies
- Recurrent airway infections
- Renal anomalies/urogenital problems\*
- Hyperextensible joints
- Lymphedema\*
- Eczema

\*only or mainly observed in deletions 22q13.3

<sup>#</sup>more common in *SHANK3* variants

#### **Acute life-threatening complications**

- Seizures
- Burning accidents due to decreased pain response
- Complications due to gastro-oesophageal reflux
- Over-heating due to hypohidrosis
- Airway infections

Further information can be obtained from the International Phelan-McDermid syndrome Foundation <https://pmsf.org> and the Consensus guidelines on Phelan-McDermid syndrome, Special Issue EJMG 2023

# PHELAN-McDERMID SYNDROME EMERGENCY CARD (Updated \_\_\_/\_\_\_/20\_\_\_)

## PERSONAL DETAILS

Name \_\_\_\_\_  
Name \_\_\_\_\_  
DOB \_\_\_\_\_ Gender \_\_\_\_\_  
Phone \_\_\_\_\_  
Address \_\_\_\_\_  
Email \_\_\_\_\_  
Phone \_\_\_\_\_

## COORDINATING PHYSICIAN DETAILS

## EMERGENCY CONTACTS

Name \_\_\_\_\_  
Relation \_\_\_\_\_  
Phone \_\_\_\_\_  
Email \_\_\_\_\_

Name \_\_\_\_\_  
Relation \_\_\_\_\_  
Phone \_\_\_\_\_  
Email \_\_\_\_\_

## Typical vital parameters of patient

Oxygen saturation (%) \_\_\_\_\_  
\_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
Breathing rate (breath/min) \_\_\_\_\_  
(Kg) \_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
Heart rate (bpm) \_\_\_\_\_  
\_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
Blood pressure (mmHg) \_\_\_\_\_  
size \_\_\_\_\_  
Temperature regulation \_\_\_\_\_  
\_\_\_\_\_

Length/height (cm)

Weight

Head circumference (cm)

( ) NG tube ( ) G-tube type and

( ) Tracheostomy ( ) Mechanical ventilation

( ) Vascular access device

## Allergies:

### Major malformations

( ) Cardiac anomaly: type \_\_\_\_\_  
Last evaluation \_\_\_/\_\_\_/20\_\_\_ surgery no/ date \_\_\_/\_\_\_/20\_\_\_  
( ) Structural brain anomaly: type \_\_\_\_\_  
Last MRI \_\_\_/\_\_\_/20\_\_\_

### Psychomotor development

( ) Normal ( ) Borderline ( ) Delayed  
( ) hypotonia, degree \_\_\_\_\_

### Cognitive development

Degree of delay: ( ) mild ( ) moderate  
( ) severe ( ) profound

### Verbal communication

( ) Absent ( ) Strongly limited ( ) Limited  
( ) Near normal ( ) Normal

### Behavioural problems

( ) Sleeping problems, type \_\_\_\_\_  
( ) Anxiety ( ) Aggression ( ) Self-injurious  
( ) Hyperactivity ( ) Autism spectrum disorder  
Likes: \_\_\_\_\_  
Dislikes: \_\_\_\_\_

### Medical complications

( ) Food intolerance: ( ) Lactose ( ) Gluten  
Other \_\_\_\_\_ Special diet \_\_\_\_\_  
( ) Gastrointestinal reflux ( ) Cyclic vomiting  
( ) Constipation ( ) Diarrhoea  
( ) Hearing loss: ( ) sensorineural ( ) conductive  
( ) mild ( ) moderate ( ) severe ( ) hearing aids  
( ) Visual impairment: type \_\_\_\_\_ ( ) glasses  
( ) Increased pain tolerance  
( ) Pneumonia (recurrent), dates \_\_\_\_\_  
( ) Ear infections (frequent) ( ) Sinus infections  
( ) Renal/genital problems: type \_\_\_\_\_  
( ) Hip problems: type \_\_\_\_\_  
( ) Lymphedema type \_\_\_\_\_  
( ) Dental anomalies: ( ) cavities ( ) crowding  
( ) allows inspection  
( ) Other medical problems: type \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

**Medical treatment**

Medication	Dosage	Frequency	Reason
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