

## 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#
- Sleeping problems#
- 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

#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 Consensus guidelines on Phelan-McDermid syndrome, <https://ern-ithaca.eu/documentation/phelan-mcdermid-guideline/>, and the Phelan-McDermid syndrome Foundation <https://pmsf.org>



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

**PERSONAL DETAILS**

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

**COORDINATING PHYSICIAN DETAILS**

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

**EMERGENCY CONTACTS**

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

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

**Typical vital parameters of patient**

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

Length/height (cm) \_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
Weight (Kg) \_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
Head circumference (cm) \_\_\_\_\_ ( \_\_\_/\_\_\_/20\_\_\_ )  
( ) NG tube ( ) G-tube type and size \_\_\_\_\_  
( ) 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
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____