**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 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 COORDINATING PHYSICIAN DETAILS**

Name \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Name\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**EMERGENCY CONTACTs**

Name\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Name\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**Typical vital parameters of patient**

Oxygen saturation (%) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Length/height (cm) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_(\_\_\_/\_\_\_/20\_\_\_\_)

Breathing rate (breath/min) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Weight (Kg)\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_(\_\_\_/\_\_\_/20\_\_\_\_)

Heart rate (bpm) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Head circumference (cm) \_\_\_\_\_\_\_\_\_\_\_(\_\_\_/\_\_\_/20\_\_\_)

Blood pressure (mmHg) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ ( ) NG tube ( ) G-tube type and size\_\_\_\_\_\_\_\_\_\_\_\_

Temperature regulation\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ ( ) 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\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**Medical treatment**

Medication Dosage Frequency Reason

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