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., 2023, this issue.

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

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

_____

_____

_____

_____

_____

_____