

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)
- 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

Less frequently occurring problems (<30%)

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](#) and the Consensus guidelines on Phelan-McDermid syndrome, [Special Issue EJMG 2023](#).

