28511  Partington-Anderson syndrome

Eponyms:

Inheritance: autosomal recessive

Semeiological Synthesis: Facio-neurologic disorder. Friendly personality, microcephaly, peculiar dysmorphic face, columnella below the nasal alae.

Group

Sub group

Signs:

NEUROLOGICAL DISORDERS

behaviour, changes

- happy nature, pleasant nature, happy disposition

brain anomalies

- microcephaly, microcrania

OROCRANIOFACIAL ANOMALIES

facies, modified appearance

- dysmorphic face

- facial dysmorphism due to cranial changes including microcephaly

forehead-orbital region, changes

- eyebrows, bush

maxilla-cheek changes

- cheeks, prominent, pouting, full, chubby, puffy

nose, modified appearance

- nose, small, nose tiny, nose short, not including very small

OTHERS

inheritance

- inheritance, autosomal recessive

PRENATAL-NEONATAL MODIFIED DATA

foetal changes

- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR

- foetal changes, recognized by ultrasound techniques

prenatal diagnosis

- prenatal diagnosis, echographic

SKELETAL DISORDERS

stature, growth, modified habitus

- growth delayed, failure to thrive, growth retardation

Super group: dysmorphic face-mental retardation

Super aggreg.

Aggregations: foetal changes, recognized by ultrasound techniques

OTHER

craniofacial dysmorphism due to cranial changes, including microcephaly
dysmorphic face

Differential diagnosis: 1630  Angelman syndrome

26810  Williams syndrome

Bibliography  OMIM ID: 260555