foetal maternal trimethadione syndrome

Eponyms:  tridione/paramethadione/paradione
          foetal effects
          troxidone embryopathy

Inheritance:  no genetic


Group  Sub group  Signs:

  AUDITORY DISORDERS
    deafness
      deafness sensorineural, including unspecified type
      deafness, in syndromic association
  
  CARDIOVASCULAR DISORDERS
    cardiac congenital defects
      cardiopathy, congenital
  
  GASTROINTESTINAL DISORDERS
    oesophageal anomalies
      oesophageal atresia, brachyoesophagus
  
  GENITAL DISORDERS
    female genitalia, modified not including ambiguity
      clitoris, hypertrophic, clitoromegaly
    male genitalia, modifications not including ambiguity
      hypospadias
  
  JOINT DISORDERS
    joint, laxity, dislocations
      hip dislocation
  
  NEUROLOGICAL DISORDERS
    behaviour, changes
      dysarthria
      brain anomalies
        microcephaly, microcrania
      mental retardation
        mental retardation
  
  OCULAR DISORDERS
    eye, motility defects
      strabismus convergent, esotropia, misalignment of the visual axes of the eyes
    eyelids, anomalies
      eyelids, ptosis
    visus defects
      myopia
  
  OROCRANIOFACIAL ANOMALIES
    external ear malformations
      ear lobe, cup shaped
      ear, low set
      helix dimple
    facies, modified appearance
      dysmorphic face
    forehead-orbital region, changes
      angioa frontalis, naevus flammeus frontalis, forehead hemangioma, angel's kiss, Bossard spot, salmon patch
      epicanathus
      eyebrows, arcuated
      frontal bossing
- synophris, eyebrows meeting across the midline

*lip-palate, cleft*
- lip and palate cleft, cheilognatopalatoschisis
  - not including isolated palate cleft

*mandibular changes*
- micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia

*maxilla-cheek changes*
- midface hypoplasia, malar hypoplasia, hypoplastic zygomata

*neck, modified appearance*
- pterygium colli, webbed neck, including:
  - thickened nuchal fold, nuchal thickening

*nose, modified appearance*
- nasal bridge, nasal root, broad, wide, squared
- nasal bridge, nasal root, low
- nose, pointed, anteverted nostrils
- nose, small, nose tiny, nose short, not including very small

*palatopharyngeal changes*
- palate cleft, palatoschisis, including submucous, not including lip and palate cleft

**OTHERS**

*inheritance*
- inheritance, no genetic

*supergroups*
- arthro-facio-neuro-oculo-oto-skeletal disorders
- arthro-neuro-oculo-oto-skeletal disorders
- arthro-neuro-skeletal disorders
- arthro-oculo-oto disorders
- cardio-facio-gastrointestinal-skeletal disorders
- cardio-facio-genito-neuro-skeletal disorders
- cardio-facio-neuro-oto-skeletal disorders
- cardio-facio-urological disorders
- cardio-genito-skeletal-urological disorders
- facio-gastrointestinal-neuro-oculo-skeletal disorders
- facio-genito-neuro-skeletal disorders
- facio-genito-oculo-oto disorders
- facio-genito-skeletal-urological disorders
- facio-urological disorders
- gastro-intestinal-oculo-skeletal disorders
- genito-oculo-oto-urological disorders
- oto-skeletal-urological disorders

**PRENATAL-NEONATAL MODIFIED DATA**

*foetal changes*
- birth length decreased; low birth length
- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
- foetal changes, induced by environmental agents not including: infections
- foetal changes, recognized by ultrasound techniques

*prenatal diagnosis*
- prenatal diagnosis, echographic

**SKELETAL DISORDERS**

*skull shape, changes*
- craniosynostosis

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spine, changes
- scoliosis, kyphoscoliosis

UROLOGICAL DISORDERS
kidney, malformations
- kidney, malformation, including horseshoe kidney

Super group:
- arthro-facio-neuro-oculo-oto-skeletal disorders
- arthro-neuro-skeletal disorders
- arthro-oculo-oto disorders
- cardio-facio-gastrointestinal-skeletal disorders
- cardio-facio-genito-neuro-skeletal disorders
- cardio-facio-neuro-oculo-oto disorders
- cardio-facio-neuro-skeletal disorders
- cardio-facio-oculo-skeletal-urological disorders
- cardio-facio-oto-skeletal disorders
- cardio-facio-urological disorders
- cardio-genito-skeletal-urological disorders
dysmorphic face-mental retardation: deafness
- facio-gastrointestinal-neuro-oculo-skeletal disorders
- facio-genito-neuro-skeletal disorders
- facio-genito-oculo-oto disorders
- facio-genito-skeletal-urological disorders
- facio-neuro-oculo-skeletal disorders
gastrointestinal-oculo-skeletal disorders
genito-oculo-oto-urological disorders
- oto-skeletal-urological disorders

Super aggreg.
Aggregations:
DEAFNESS
deafness, in syndromic association

FOETAL CHANGES
foetal changes, induced by environmental agents not including infections
foetal changes, recognized by ultrasound techniques

OTHER
craniofacial dysmorphism due to cranial changes, including microcephaly
dysmorphic face

Differential diagnosis:
- 9580 foetal maternal toluene syndrome
- 28097 Sao Paulo syndrome
- 28435 Wolff syndrome

Bibliography
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