

## Opis choroby \*

### Definicja

An extremely rare type of arthrogryposis multiplex congenita characterized by the combination of multiple joint contractures with movement limitation, microstomia with a whistling appearance of the mouth that may cause feeding, swallowing, and speech difficulties, a distinctive expressionless facies, severe developmental delay, central and autonomous nervous system dysfunction (excessive salivation, temperature instability, myoclonic epileptic fits, bradycardia), occasionally Pierre-Robin sequence, and lethality generally occurring during the first months of life. Arthrogryposis multiplex congenita-whistling face syndrome has been suggested to be a fetal akinesia deformation sequence.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych IIIum syndrome	Zespół IIIum

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
1150	208155	Q87.8

**Kod ICD11**  
LD26.41

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### \*Źródło

orphanet