

## Opis choroby \*

### Definicja

A rare genetic syndrome characterized by skeletal anomalies, including short stature, ridging of the metopic suture, a fusion of cervical vertebrae, thoracic hemivertebrae, scoliosis, sacral hypoplasia, short middle phalanges. Patients also had a moderate intellectual disability and abducens palsies. Glucose intolerance and imperforate anus were also described.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Christian syndrome
	Zespół Christiana

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
1436	309620	Q87.5

### Kod ICD11

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### \*Źródło

orphanet