

## Opis choroby \*

### Definicja

Joubert syndrome with ocular defect is, along with pure JS, the most frequent subtype of Joubert syndrome and related disorders (JSRD, see these terms) characterized by the neurological features of JS associated with retinal dystrophy.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych JS-O	JS-O
	Zespół Joubert z retinopatią
	Joubert syndrome with retinopathy

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
220493	617121	Q04.3

**Kod ICD11**  
LD20.0Y

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### \*Źródło

orphanet