

## Opis choroby \*

### Definicja

Stimmler syndrome is characterised by the association of microcephaly, low birth weight and severe intellectual deficit with dwarfism, small teeth and diabetes mellitus. Two cases have been described. Biochemical tests reveal the presence of high levels of alanine in the urine and elevated alanine, pyruvate and lactate levels in the blood.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

3199

#### Kod OMIM

202900

#### Kod ICD10

Q87.1

#### Kod ICD11

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### \*Źródło

orphanet