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
-

---

*Źródło

orphanet