

Opis choroby *

Definicja

Congenital muscular dystrophy-infantile cataract-hypogonadism syndrome is characterized by congenital muscular dystrophy, infantile cataract and hypogonadism. It has been described in seven individuals from an isolated Norwegian village and in one unrelated individual. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja

Choroba

Synonimy

Bassoe syndrome

Zespół Bassoe

Kod ORPHA

1875

Kod OMIM

254000

Kod ICD10

G71.0

Kod ICD11

8C70.6

*Źródło

orphanet