

Opis choroby *

Definicja

LIG4 syndrome is a hereditary disorder associated with impaired DNA double-strand break repair mechanisms and characterized by microcephaly, unusual facial features, growth and developmental delay, skin anomalies, and pancytopenia, which is associated with combined immunodeficiency (CID).

Dane

Klasyfikacja

Choroba

Synonimy

DNA ligase IV deficiency
Niedobór ligazy DNA IV
Zespół ligazy 4
Ligase 4 syndrome

Kod ORPHA

99812

Kod OMIM

606593

Kod ICD10

D81.1

Kod ICD11

4A01.1Y

*Źródło

orphanet