

Opis choroby *

Definicja

Spondyloepimetaphyseal dysplasia congenita, Shohat type is characterized by severely disproportionate short stature, short limbs, small chest, short neck, thin lips, severe lumbar lordosis, marked genu varum, joint laxity, distended abdomen, mild hepatomegaly and splenomegaly.

Dane

Klasyfikacja

Choroba

Synonimy

SEMD, Shohat type

SEMD, typ Shohat

Kod ORPHA

93352

Kod OMIM

602557

Kod ICD10

Q77.7

Kod ICD11

LD24.3

*Źródło

orphanet