

Opis choroby *

Definicja

Camptodactyly-tall stature-scoliosis-hearing loss syndrome is characterised by camptodactyly, tall stature, scoliosis, and hearing loss (CATSHL). It has been described in around 30 individuals from seven generations of the same family. The syndrome is caused by a missense mutation in the FGFR3 gene, leading to a partial loss of function of the encoded protein, which is a negative regulator of bone growth.

Dane

Klasyfikacja

Choroba

Synonimy

CATSHL syndrome

Zespół CATSHL

Camptodactyly-tall stature-scoliosis-deafness syndrome

Kod ORPHA

85164

Kod OMIM

610474

Kod ICD10

Q87.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet