DISEASE:
Camptodactyly-tall stature-scoliosis-hearing loss syndrome

NAME:	Camptodactyly-tall stature-scoliosis-hearing loss syndrome
DESCRIPTION:	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.
ORPHACODE:	85164
SYNONYMS:	CATSHL syndrome Camptodactyly-tall stature-scoliosis-deafness syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FGFR3
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RELATED CONTENT

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- Short Stature (gene panel)

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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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