DISEASE:
Björnstad syndrome

NAME:	Björnstad syndrome
DESCRIPTION:	Björnstad syndrome is characterized by congenital sensorineural hearing loss and pili torti.
ORPHACODE:	123
SYNONYMS:	Deafness-pili torti-hypogonadism syndrome Hearing loss-pili torti-hypogonadism syndrome
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	BCS1L
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Mitochondrial complex III deficiency / Bjornstad syndrome / Gracile syndrome
- Mitochondrial disorders (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone

Related Gene Panels

- mitochondrial disease, nuclear based (343 genes) - VUB