DISEASE:
Schilbach-Rott syndrome

NAME:	Schilbach-Rott syndrome
DESCRIPTION:	Schilbach-Rott syndrome (SRS) is an autosomal dominant dysmorphic disorder that is characterized by dysmorphic facies with hypotelorism, blepharophimosis, and cleft palate, and the frequent occurrence of hypospadias in males.
ORPHACODE:	2353
SYNONYMS:	BRSS Hypotelorism-cleft palate-hypospadias syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PTCH1
CREATED:	04 Feb 2020 - 15:13
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB

Related Analytes

- patched 1