sciensano **Belgian Genetic Tests database**

DISEASE:
Intellectual disability-expressive aphasia-facial dysmorphism syndrome

NAME:	Intellectual disability-expressive aphasia-facial dysmorphism syndrome
DESCRIPTION:	A rare genetic syndromic intellectual disability characterized by moderate to severe intellectual deficiency, language deficit (completely absent or significantly impaired speech), and distinctive facial dysmorphism (long face, straight eyebrows, and, less frequently, low-set ears and café-au-lait spots). Additional, variably observed features include motor delays, behavioral difficulties, and seizures.
ORPHACODE:	436151
SYNONYMS:	Intellectual disability-loss of expressive language-facial dysmorphism syndrome
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	SETBP1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2412

RELATED CONTENT

Related Genetic Tests

- Schinzel-Giedion midface retraction syndrome

Related Laboratories

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

Related Analytes

- SET binding protein 1