DISEASE:
Aniridia-cerebellar ataxia-intellectual disability syndrome

NAME:	Aniridia-cerebellar ataxia-intellectual disability syndrome
DESCRIPTION:	A rare, congenital, neurological disorder characterized by the association of partial bilateral aniridia with non-progressive cerebellar ataxia, and intellectual disability.
ORPHACODE:	1065
SYNONYMS:	Gillespie syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ITPR1 PAX6
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Aniridia

Related Laboratories

- Centrum Medische Genetica - UZ Gent

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- inositol 1,4,5-trisphosphate receptor type 1
- paired box 6