DISEASE:
MIRAGE syndrome

NAME:	MIRAGE syndrome
DESCRIPTION:	A rare genetic disease characterized by pre- and postnatal growth restriction, developmental delay, adrenal hypoplasia, genital abnormalities (such as microphallus, hypospadias, or cryptorchidism), thrombocytopenia and/or anemia, recurrent severe invasive infections, and enteropathy with chronic diarrhea. Myelodysplastic syndrome and dysmorphic features (including downslanting palpebral fissures, low-set and posteriorly rotated ears, anteverted nares, camptodactyly, and arachnodactyly, among others) may also be observed.
ORPHACODE:	494433
SYNONYMS:	Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital anomalies-enteropathy syndrome Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital phenotypes-enteropathy syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SAMD9
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RELATED CONTENT

Related Genetic Tests

- Myeloid/lymphoid neoplasms with germline predisposition (gene panel)
- Primary immune deficiencies (gene panel)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- sterile alpha motif domain containing 9

Related Gene Panels

- Hematologic Familiar Forms - ULG
- Primary immune deficiencies (444 genes) - KUL