DISEASE:
Generalized pseudohypoaldosteronism type 1

NAME:	Generalized pseudohypoaldosteronism type 1
DESCRIPTION:	A severe form of pseudohypoaldosteronism type 1 characterized by salt wasting in multiple organs including the kidney, colon, and sweat and salivary glands. Presentation is in the first few weeks of life with severe dehydration, vomiting and failure to thrive in association with hyponatremia, hyperkalemia and metabolic acidosis as well as elevated aldosterone and renin levels. No remission is reported and patients suffer from recurrent life-threatening episodes of salt loss.
ORPHACODE:	171876
SYNONYMS:	Autosomal recessive PHA1 Autosomal recessive pseudohypoaldosteronism type 1 Generalized PHA1
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SCNN1A SCNN1B SCNN1G
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/424

RELATED CONTENT

Related Genetic Tests

- Bronchiectasies with or without elevated sweat chloride panel (5 genes)
- Tubulopathy (gene panel)

Related Laboratories

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

Related Analytes

- sodium channel epithelial 1 subunit alpha
- sodium channel epithelial 1 subunit beta
- sodium channel epithelial 1 subunit gamma

Related Gene Panels

- Cystic Fibrosis / Liddle syndrome / Pseudohypoaldosteronism type 1 (3 genes) - ULG
- Pulmonary/Bronchiectasies (5 genes) - IPG
- Tubulopathy/Nephrolithiasis (106 genes) - IPG