

**DISEASE:**  
**Renal pseudohypoaldosteronism type 1**

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| <b>NAME:</b>        | Renal pseudohypoaldosteronism type 1                                                                                                                                                                                                                                                                                                                                                                             |
| <b>DESCRIPTION:</b> | A form of pseudohypoaldosteronism type 1 characterized by mild mineralocorticoid resistance that is restricted to the kidneys and that usually improves in early childhood. Typical presentation is in the neonatal period with weight loss, failure to thrive, vomiting and dehydration in association with hyponatremia, hyperkalemia and metabolic acidosis as well as elevated aldosterone and renin levels. |
| <b>ORPHACODE:</b>   | 171871                                                                                                                                                                                                                                                                                                                                                                                                           |
| <b>SYNONYMS:</b>    | Autosomal dominant PHA1<br>Autosomal dominant pseudohypoaldosteronism type 1<br>Renal PHA1                                                                                                                                                                                                                                                                                                                       |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                                                                                                                                                                  |
| <b>ANALYTE(S):</b>  | <u>NR3C2</u>                                                                                                                                                                                                                                                                                                                                                                                                     |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                                                              |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                                                              |

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