Pseudohypoaldosteronism

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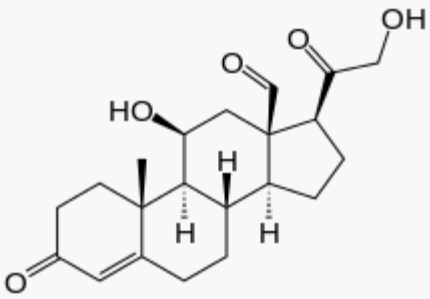
Pseudohypoaldosteronism (PHA) is a condition that mimics hypoaldosteronism.[1] However, the condition is due to a failure of *response* to aldosterone, and levels of aldosterone are actually elevated, due to a lack of feedback inhibition.

This syndrome was first described by Cheek and Perry in 1958.[2] Later pediatric endocrinologist Aaron Hanukoglu reported that there are two independent forms of PHA with different inheritance patterns: Renal form with autosomal dominant inheritance exhibiting salt loss mainly from the kidneys, and multi-system form with autosomal recessive form exhibiting salt loss from kidney, lung, and sweat and salivary glands.[3] [4]

Pseudohypoaldosteronism



In pseudohypoaldosteronism, aldosterone is elevated (hyperaldosteronism), but because the body fails to respond to it, it appears similar to hypoaldosteronism.

Classification and external resources

OMIM	177735 614495 614491 614496 614492 145260 264350 177735 614495 614491 614496 614492 145260
DiseasesDB	= [http://apps.who.int/classifications/icd10/browse/2016/en#/N25.8 N25.8.htm ICD10 = N25.8]
eMedicine	article/924100
MeSH	D011546

[edit on Wikidata]

Treatment of severe forms of PHA requires relatively large amounts of sodium chloride.[5] These conditions also involve hyperkalemia.[6]

Types include:

Type	OMIM	Gene	Description
PHA1AD	177735	MLR	with sodium wasting
PHA1AR	264350	SCNN1A, SCNN1B, SCNN1G of the epithelial sodium channel	with sodium wasting
PHA2	145260	WNK4, WNK1	without sodium wasting. TRPV6 may be involved.[7]

References [edit]

1. ^ "Pseudohypoaldosteronism: Overview - eMedicine Pediatrics: General Medicine". Retrieved 2009-03-06.
2. ^ Boyle WA, Nerbonne JM (Apr 1991). "A novel type of depolarization-activated K+ current in isolated adult rat atrial myocytes". *The American Journal of Physiology* **260** (4 Pt 2): H1236–47. doi:10.1136/adc.33.169.252. PMC 2012226. PMID 13545877.
3. ^ Hanukoglu A (Nov 1991). "Type I pseudohypoaldosteronism includes two clinically and genetically distinct

entities with either renal or multiple target organ defects". *The Journal of Clinical Endocrinology and Metabolism* **73** (5): 936–44. doi:10.1210/jcem-73-5-936. PMID 1939532.
4. ^ Hanukoglu I, Hanukoglu A (Jan 2016). "Epithelial sodium channel (ENaC) family: Phylogeny, structure-function, tissue distribution, and associated inherited diseases.". *Gene* **579** (2): 95–132. doi:10.1016/j.gene.2015.12.061. PMID 26772908.
5. ^ Hanukoglu A, Hanukoglu I (2010). "Clinical improvement in patients with autosomal recessive pseudohypoaldosteronism and the necessity for salt supplementation.". *Clinical and Experimental Nephrology* **14** (5): 518–519. doi:10.1007/s10157-010-0326-8. PMID 20661616.
6. ^ Pseudohypoaldosteronism at the US National Library of Medicine Medical Subject Headings (MeSH)
7. ^ Yang SS, Hsu YJ, Chiga M, Rai T, Sasaki S, Uchida S, Lin SH (Apr 2010). "Mechanisms for hypercalciuria in pseudohypoaldosteronism type II-causing WNK4 knock-in mice". *Endocrinology* **151** (4): 1829–36. doi:10.1210/en.2009-0951. PMID 20181799.

External links [edit]

- GeneReviews/NCBI/NIH/UW entry on Pseudohypoaldosteronism Type II
- Pseudohypoaldosteronism support page on Facebook

See also [edit]

- Hyperchloremic acidosis
- Pseudohyperaldosteronism

v · t · e ·	**Diseases of the endocrine system (E00–E35, 240–259)**	[show]
v · t · e ·	**Diseases of the urinary system (N00–N39, 580–599)**	[show]
v · t · e ·	**Genetic disorder, protein biosynthesis: Transcription factor/coregulator deficiencies**	[show]
v · t · e ·	**Diseases of ion channels**	[show]
v · t · e ·	**Deficiencies of intracellular signaling peptides and proteins**	[show]

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