Pseudohypoaldosteronism, Type IIA

Alternative Names
PHA2A
Hyperpotassemia and Hypertension, Familial
Hypertensive Hyperkalemia, Familial
Gordon Hyperkalemia-Hypertension Syndrome

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic diseases > Metabolic disorders

Incidence per 100,000 Live Births
Unknown

OMIM Number
145260

Mode of Inheritance
Autosomal Dominant and Autosomal Recessive

Gene Map Locus
1q31-q42

Description
Pseudohypoaldosteronism (PHA) is assigned to a heterogeneous group of disorders of electrolyte metabolism which is distinguished by a clear state of renal tubular unresponsiveness or resistance to the action of aldosterone. The condition is described by hyperkalemia, metabolic acidosis and normal glomerular filtration rate (GFR). Several features are observed in this condition such as volume depletion or hypervolemia, renal salt wasting or retention, hypotension or hypertension, and elevate, normal or low levels of rennin and aldosterone.

Molecular Genetics
The molecular basis for patients suffering from PHA-II is connected to mutations in WNK1 or WNK4. WNKs include a family of serine-threonine protein kinases with unusual placement of the catalytic lysine compared to all other protein kinases. The function of WNK1 and WNK4 lies in regulating chloride co-transporters of the distal nephron and further epithelia.

Epidemiology in the Arab World
Kuwait
Pinto et al. (2003) reported a case of osteopetrosis that was associated with hyperkalemia on admission to Al-Adan Hospital. He was found later to suffer from pseudohypoaldosteronism (PHA). The patient was a first born child to consanguineous parents with 34 weeks gestation and birth weight of 1.865kg. The patients' extended family of both parents revealed no family history of a similar condition. The subject suffered from hyperkalemia, high serum aldosterone and rennin levels, normal cortisol and 17-OH progesterone, renal salt loss and normal sweat electrolytes, and all of these features were found to be compatible with pseudohypoaldosteronism (PHA) type 1. The case was found to have the autosomal dominant (AD) form of PHA type 1, including only the renal tubules. Pinto et al. (2003) suggested this to be a new mutation since both parents were tested normal for electrolytes.

References
Pinto R, John I, Qabazard Z. Osteopetrosis associated with hyperkalaemia: a case report. Kuwait Med J. 2003; 35(3): 216-8.

Related CTGA Records
N/A

External Links
http://www.ncbi.nlm.nih.gov/omim/145260
http://emedicine.medscape.com/article/924100-overview

Contributors
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