Reviewer’s report

Title: Isolated hypoaldosteronism as first sign of X-linked Adrenal Hypoplasia Congenita caused by a novel mutation in NR0B1/DAX-1 gene: a case report.

Version: 0 Date: 05 Apr 2019

Reviewer: Ece Bober

Reviewer's report:

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I don't think of this case has unusual onset of AHC that started with isolated hypoaldosteronism as first and only sign of disease. The case was admitted to hospital for failure to thrive that is the symptom of hypocortisolism. Blood glucose level of the case was not given (hypoglycemia?). Normal plasma cortisol level doesn't rule out hypocortisolism whenever ACTH level was high. Mild pigmentation of the external genitalia was also the sign of hypocortisolism.

The short Synacthen test was unnecessary in this case because of the high ACTH levels.

There are many similar cases of AHC that salt-wasting is the first presentation and diagnosed to have isolated primary hypoaldosteronism mistakenly (1,2).

In a study from Italy, authors investigated the data of newborns and infants that presented with hyponatremia in a single center during ten years. They found 51 infants, 19 of them diagnosed as CAH, and four of them diagnosed as AHC (2).

Table 2 is unnecessary. There are more than 200 NR0B1/DAX-1 gene mutation cases in the literature.

Discussion is successful and comprehensive to understanding the newborn mineralocorticoid regulation.

This case, contains no new knowledge about AHC, except the NR0B1/DAX-gene mutation was novel.


**Are the methods appropriate and well described?**
If not, please specify what is required in your comments to the authors.

Yes

**Does the work include the necessary controls?**
If not, please specify which controls are required in your comments to the authors.

No

**Are the conclusions drawn adequately supported by the data shown?**
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Yes

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Not relevant to this manuscript

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