Reviewer’s report

Title: A Case Report: Familial Glucocorticoid Deficiency associated with Familial Focal Segmental Glomerulosclerosis.

Version: 1  Date: 13 September 2012

Reviewer: Karen Heath

Reviewer’s report:

The authors report on an 8-month old boy who presents with isolated glucocorticoid deficiency who later develops focal segmental glomerulosclerosis. The case is precisely documented and is of interest to clinicians in this field. It is a shame that molecular analysis, through a collaboration, was not undertaken as the identification of the mutation and implicated gene would add the missing piece to the puzzle.

Major compulsory revisions
None

Minor essential revisions
In the discussion the genes need to be written in italics.

In the discussion they state that FGD is caused by two genes but they should discuss that FGD cases are caused by mutations in one of four genes: MC2R, MRAP and STAR, encoding components of the ACTH signaling-steriodogenic pathway, and NNT which encodes an integral protein of the inner mitochondrial membrane and which appears to be of primary importance for ROS detoxification in adrenocortical cells (Meimaridou et al, Nat Genet 2012:44:740-2).

Discretionary revision
English editing of various phrases as indicated below.

Case presentation
First paragraph – Change the sentence “cortisol was low with very much raised ACTH levels” to “cortisol was low with highly elevated ACTH levels”

Third paragraph change sentence “Renal biopsy done” to “Renal biopsy was undertaken”

Discussion
First paragraph, last sentence needs to be clear, for example – end with “and high ACTH. The relatives come from a family with a history of cousin marriages.”

In the sentence starting “He was found to have FGD type 2 with positive molecular genetics for MRAP gene mutation”, I would rewrite to “He was molecularly confirmed to have FGD type 2.”
In the next sentence need to correct “nine month” to “nine months”.

In the 3rd paragraph of the discussion should correct “suggest genetic link” to “suggested a genetic link”.

Editing - references cited in text should have last name but not initials.

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Needs some language corrections before being published

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**

'I declare that I have no competing interests' below.