Reviewer's report

Title: Hypospadias in an Egyptian infant with X linked congenital adrenal hypoplasia: a possible new syndrome.

Version: 1 Date: 31 August 2012

Reviewer: Tsutomu Ogata

Which of the following best describes what type of case report this is?: New associations or variations in disease processes

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is the case report ethical?: No

Is there any missing information that you think must be added before publication?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Is the anonymity of the patient protected?: Yes

Comments to authors:

This manuscript describes an interesting association of DAX1 mutation and CXorf6 mutation.

I have several comments that hopefully help to improve this manuscript.

1. The word [syndrome] must be deleted throughout the manuscript. This boy has an association of DAX1 mutation and CXorf6 mutation, rather than a syndrome.

2. The word [CXorf6] must be replaced with [MAMLD1]. MAMLD1 is the formal name for this gene.

3. ARG327PRO should be rewritten as [p.Arg327Pro], and 325delG should be rewritten as c.325delG.
4. Do you have some data showing that p.Arg327Pro is a pathologic mutation rather than a polymorphism.
5. Figure should be reduced in number. Fig. 3 only would be enough.
6. English and typing are both poor.
7. Please describe about ethical approval.

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

**Declaration of competing interests:**

I have nothing to declare.