Reviewer's report

Title: Bardet-Biedl Syndrome with end stage kidney disease at an early age: a case report

Version: 2 Date: 24 March 2010

Reviewer: Marie Cassart

Which of the following best describes what type of case report this is?: Unexpected or unusual presentations of a disease

Has the case been reported coherently?: No

Is the case report authentic?: Yes

Is the case report ethical?: Yes

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?: No

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: No

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

Is the anonymity of the patient protected?: Yes

Comments to authors:

Dear authors,

Major comments

Bardet-Biedl (BB) is a rare autosomal recessive disorder characterized by multisystemic anomalies. The definite diagnosis is often delayed (excepted in cases of recurrence) due to late appearance of the symptoms. The interest of the present case is the unusual early occurrence of renal insufficiency associated with poor prognosis. This is the main point of interest on which the report should be focused. The report should therefore be shortened and rewritten focusing on its peculiarity.

Meanwhile, it is surprising that the diagnosis has not been established earlier in a
context of recurrence (elder sister affected) and consanguinity of the parents. The antenatal data are not provided (did the mother had US during her pregnancy? The fetal kidneys in BB have a typical pattern and polydactyly can be detected. This should be mentioned.

More over, the absence of genetic definite diagnosis weakens the paper as it is an unusual presentation of BB disorder. It suggests the possibility of another syndromal association. This should be discussed.

Minor comments
There are too many figures without interest.
I would add a genetic tree of the family outlining affected persons.
The US image of the kidney is typical of BB in early childhood. It would be interesting to give the age of the child at this US (we published in 2004 an article on the sonographic aspects of the kidneys in fetuses and children).

Best regards
Marie Cassart

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

**Declaration of competing interests:**

'I declare that I have no competing interests'