Reviewer's report

Title: Coffin Siris Syndrome with Mayer-Rokitansky-Kuster-Hauser syndrome

Version: 2 Date: 15 March 2010

Reviewer: Indraneel Banerjee

Which of the following 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?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

Interesting combination of 2 unusual conditions. Both syndromes have been well reported previously. However, the combination has not been reported in the literature and there may be clues to the aetiology of both conditions. The manuscript should be rearranged to reflect the strength of the unusual association, not simply a description of the Coffin-Sirris syndrome.

Child's height is provided with a centile measurement, but not specified which reference standard was used. Similarly, parents height should be recorded with midparental height centile to define the degree of short stature.

Hirsutism has been mentioned, which can be associated with the MRKH syndrome. If possible, androgen levels could be provided.

It is not clear why the child had MR scans, particularly of the abdomen. This
should be justified, particularly from an ethical perspective.

A number of discussion points are irrelevant, for example the statement that Coffin-Siris syndrome arises from new mutations, when no genetic mutations have been identified. The order of description of CS and MRKH should be revised.

Authors should try and piece together the links between 2 unusual syndromes, for e.g mesodermal developmental disorders, providing insight into the causal aetiology and possible gene candidates.

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

**Declaration of competing interests:**

I declare that I have no competing interests.