Reviewer’s report

Title: A case of Cornelia de Lange syndrome from Sudan

Version: 2 Date: 10 September 2006

Reviewer: Raoul C Hennekam

Reviewer’s report:

General

The goal of this paper is to show a patient with CDLS and an ethnic background that has not been published before. The paper should put emphasis on this goal, and not on other aspects.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

a. The wordings used to describe the patient in general are good. We need more information on growth (birth weight, length at time of investigations, and also weight and skull circumference). The clinical pictures however have an insufficient resolution, which should absolutely be ameliorated as otherwise colleagues will not recognise it. Figure 1b (the overview) is a useful picture. Fig 1a (the side view) can better be combined with fig 2d (face en face), making 2d a new number 2a, and 1a the new number 2b. For the limbs the present figs 2a and 2b can be added as 2c and 2d, prvided the figures are rotated in such a way the fingers point upwards. Fig 2c and fig 3 do not add, and can both be deleted. The authors describe an unusual feature crowded ribs. I have never seen this in the large number of patients with CDLS in the past, so it would be justified if the authors provide a picture of the radiograph (a-p thorax) to show this.

b. The goal is describing a patient with an ethnic background that has not been described before. So that should get all attention. I suggest to delete the whole paragraph on the cytogenetic abnormalities, as this is out of date (even the patient described by Ireland et al [your ref 16] has been found to have a NIPBL mutation, so the 3q abnormality was really coincidense). This paragraph starts at the end of page 3 and goes on until page 6 upper part. Just referral to the DeScipio paper (your ref 18) is sufficient as this reviews all chromosome anomalies is is easy accessible to all readers. The part on the molecular findings however is insufficient and sometimes does not pay sufficient attention to the work of various groups. The gene NIPBL has been found by two groups at the same time (your refs 3 and 20) and both should be cited side-by-side. Th fact that another gene has been described which may give rise to a possible X-linked form of CDLS (your ref 21) is not mentioned sufficiently re the X-linked pattern of inheritance; this hould be added

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

a. The present patient is described to have hypotelorism; this is not evident on the pictures provided, in which the distance between the palpebral fissures seems normal. I do not understand why the authors state that failure to thrive is no symptom in CDLS, as growth failure both in length and in weight is universal in CDLS; this should be removed. The authors describe the skin as thin. However, it seems they mean that the patient has decreased subcutaneous fat tissue secondary to the feeding problems (as was the large liver). The statement of the skin s therefore no symptom of CDLS as such in this patient, and should not be mentioned in the abstract, but only once as a symptom fitting the malnutrition

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Discretionary Revisions (which the author can choose to ignore)

the authors may wish to add the recent other series of larger groups of Western World patients, to substantiate that a mutation in NIPBL can be found in about 43% of cases in total. The other publications can be easily found through a pubmed search, and data are already available in the abstracts, which will be available too.

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions
Level of interest: An article of limited interest

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:

I declare that I have no competing interests