Reviewer’s report

Title: Identification of a novel de novo mutation in the NIPBL gene in an Iranian patient with Cornelia de Lange Syndrome: a case report

Version: 4 Date: 29 March 2010

Reviewer: I Barisic

Which of the following best describes what type of case report this is?: Findings that shed new light on the possible pathogenesis of a disease or an adverse effect

If other, please specify:
indentification of the new disease causing mutation

Has the case been reported coherently?: Yes

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?: No

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:

1) General comments
The authors report on a case of CdLs and identify a novel disease causing mutation in the NIPBL gene.

2) Revisions necessary for publication.

Abstract
Introduction

line 4 - estimated prevalence as high as 1/10 000.

Recently it was proven that this old estimate made by Opitz (1985) is highly exaggerated and not based on the actual research results. Population-based study estimated prevalence for mild and classical CdLs to be 1.6 - 2.2 per 100.000 births (ref. 7). Line 14 - there has been a mixing up with references. Ref. 7 is not referring to sister chromatid cohesion, but ref. 8. Please, check all ref.

Case presentation

line 8 - cryptorchidism instead of criptoychridism line 13 - length instead of height

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

Declaration of competing interests:

'I declare that I have no competing interests'