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: 7 Date: 13 November 2010

Reviewer: Matt Deardorff

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?: 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:

Comments

The authors report a patient with classical CdLS caused by a novel mutation in exon 10 of NIPBL. It is succinctly written and easily understood and provides a useful and accurate brief review of the field.

Abstract

Use of the phrase “are responsible for” implies that mutations in the known genes explain all of CdLS. Perhaps “have been identified in” might be more clear.

The mutation “presumably” results in a premature termination since the authors do not show its effect on expression the RNA or protein.
Figures
Because of the utility of the facial features in the diagnosis of CdLS I would strongly encourage the authors to seek permission to present a facial photograph that does not obscure the eyes. In addition, if clearer photos are available of the hands, they would help to better illustrate and educate the reader of the classic features demonstrated in CdLS.

Corrections
Abstract
Background:
The X-linked gene in CdLS is SMC1A, not SLC1.

Introduction
NIPBL is mistyped as NLPBL in the last sentence.

Quality of written English: Acceptable

Declaration of competing interests:
I declare that I have no competing interest