Reviewer's report

Title: Marie and Sainton disease - A case report

Version: 2 Date: 5 November 2010

Reviewer: Blanca Silvia González-López

Which of the following following best describes what type of case report this is?: Other

If other, please specify:

This is an interesting report. The case are illustrated well high quality photographs and radiographs.

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:

P1. Change title, Marie and Sainton disease, Cleidocranial displasia according to recommended nomenclature and classification of syndromes. (International workshop1975)

P2 Abstract
Delete from Introduction; “in the present case, chromosomal analysis and gene mapping revealed normal”. As this information correspond to case presentation.
Abstract case presentation
Delete with retained deciduous teeth and multiple impacted supernumerary teeth.

P2 Here you should be described the purpose of this article

P3 Case report
Delete named Soumyasree

Case report
Change Management to Management

P3 Discussion
Change “Cleidocranial dysplasia is a well defined clinical phenotype arising from
deregulation of intramembranous and endochondral ossification”.
To
Cleidocranial dysplasia is a well defined clinical phenotype arising from
deregulation of intramembranous and endochondral ossification due to a
mutation in Cbfa1 (Core binding factor); that affected the osteoblast-specific
transcription factor.

P3 feature change features
P3 shoulder change shoulders

P3 Delete which can be approximated in front of the chest.

P3 Change “… to confirm the diagnosis and in our case, the pathognomonic
features like absence of clavicles, broad skull sutures, numerous impacted teeth
and supernumerary teeth were present”.
To
Abnormal clavicles, wormian bones and supernumerary teeth see to be
consistent features of CCD.10 …….” in our case, the pathognomonic features
like absence of clavicles, broad skull sutures, numerous impacted teeth and
supernumerary teeth were observed”

Delete

P3…. and the phenotype shows abnormal clavicles, wormian bones and
supernumerary teeth.10 As I suggested to change this paragraph

P3 Change Mendoza- Londono R , Lee B change Mendoza- Londono and Lee

P3 Change ……”suggested that 70% of the patients were found to have point
mutation involving RUNX2, 13% due to deletion but the remaining 17% were due
to unknown reason.9
To Mendoza-Londono and Lee suggested that 70% of the CCD patients have point mutation involving RUNX2 and 13% deletion. Also Lee et al suggested that in patients whose mutation is not found by traditional sequencing, the deletion/duplication assay, either RT-qPCR/MLPA, needs to be done.

P3 Delete this paragraph as I suggested change it to the initial paragraph of the discussion

“There is a mutation in Cbfa1 (Core binding factor); the affected molecule is the transcription factor”

P3 Change “In this case, we ascertained that the etiology was unknown as gene mapping revealed a normal female karyotype 46XX.

To

Thought it has been reported that individuals with CCD could have cytogenetically visible complex chromosome rearrangements [Purandare et al 2008], in this case chromosomal analysis and gene mapping revealed normal.

In order to identify mutations in the RUNX2, molecular genetic analysis is recommended.

P4 Can change may

If you decided modify your manuscript according my suggestions these are the references that I mentioned in it.

De novo three-way chromosome translocation 46,XY,t(4;6;21)(p16;p21.1;q21) in a male with cleidocranial dysplasia

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doi: 10.1002/ajmg.a.31750
PMCID: PMC2663417

Intragenic microdeletion of RUNX2 is a novel mechanism for cleidocranial dysplasia
Quality of written English: Acceptable

Declaration of competing interests:

I declare that I have no competing interests' below