Reviewer's report

Title: 11q23 deletion syndrome (Jacobsen syndrome) with severe bleeding: a case report

Version: 0 Date: 20 Oct 2017

Reviewer: T Mattina

Reviewer's report:

1. Do you believe the case report is authentic?

Yes/No

yes

2. Do you have any ethical concerns? Please consider if local Institutional Review Board approval or ethical approval was obtained (if appropriate) and if the patient (or their parent or guardian in the case of children under 18) gave written, informed consent to publish this case and any accompanying images. A statement to this effect should appear in the manuscript.

Comments:

no ethical concerns

3. Does the Introduction explain the relevance of the case to the medical literature?

Yes/No

yes

4. Does the article report the following information? Where information is missing, please specify.

   a. The relevant patient information, including:

      - De-identified demographic information (age, gender, ethnicity)
      - Main symptoms of the patient
      - Medical, family and psychosocial history
      - Relevant past interventions and their outcomes
b. The relevant physical examination findings

yes

c. Important dates and times in this case (if appropriate, organized as a timeline via a figure or table); if specific dates could lead to patient identification, consider including time relevant to initial presentation, i.e. initial presentation at $T = 0$, follow up at $T = 1$ month.

Yes

d. Diagnostic assessments, including:
   - Diagnostic methods
   - Challenges (e.g., financial, language/cultural)
   - Reasoning and prognostic characteristics (e.g., staging), where applicable

Yes

e. Types and mechanism of intervention

yes

f. A summary of the clinical course of all follow-up visits

yes
Comments:

no comment

5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented?

Comments:

yes
6. Is the anonymity of the patient protected? Please consider any identifying information in images such as facial features or nametags, whether the patient is named etc. If not, please detail below.

Yes/No

Yes

7. Is the Abstract representative of the case presented?

Comments:

yes

8. Does the case represent a useful contribution to the medical literature?

Comments:

yes

9. Additional comments for the author(s)?

The authors report on a patient with 11q deletion detected prenatally. The patient had the Jacobsen Syndrome phenotype, including the Paris Trousseau thrombocytopenia/thrombocytopeny. Bleeding occurring in the neonatal period was more severe than expected on the basis of thrombocytopenia. The authors suggest special care when dealing with newborn patients with JS as the platelets count is not enough to establish the risk of bleeding.

I would add that the same is true in children and adults with JS.

The case has been studied in details and the report is complete and useful.
Observations:

Abstract:

Introduction: 11q23 deletion syndrome, previously known as Jacobsen syndrome, is characterized by growth retardation, psychomotor retardation, facial dysmorphism, and thrombocytopenia.

Should be:

Introduction: 11q23 deletion syndrome, also known as Jacobsen syndrome, is characterized by growth retardation, psychomotor retardation, facial dysmorphism, multiple congenital abnormalities and thrombocytopenia.

Conclusions:

Our patient with FLI1 deletion avoided massive bleeding and serious sequela by careful management after prenatal diagnosis.

Should be:

"Our patient with 11q deletion including the FLI1 gene avoided massive bleeding and serious sequela by careful"

Introduction

Introduction

64 A contiguous gene syndrome, 11q23 deletion syndrome is characterized by growth retardation, psychomotor retardation, facial dysmorphism, abnormal platelet function, and thrombocytopenia. Until the genetic region for the syndrome was found, the condition was known as Jacobsen syndrome and considered to be the same as Paris-Trousseau syndrome, which is accompanied by congenital thrombocytopenia [1].
Should be:

"A contiguous gene syndrome, 11q23 deletion syndrome, is characterized by growth retardation, psychomotor retardation, facial dysmorphism, multiple congenital abnormalities, abnormal platelet function, and thrombocytopenia. The syndrome is also known as Jacobsen syndrome and congenital thrombocytopenia/thrombocytopenia observed in most affected patients is considered to be the same as Paris-Trousseau syndrome [1]."

87 chromosome 11: 46,XY, add (11) (q23) was found.

unless the deletion is a consequence of a translocation and there is part of an unknown chromosome attached to the deleted chromosome 11q

Should be: "46,XY, del(11) (q23->qter)"

95 He exhibited skull deformities, facial asymmetry, low-set ears, inguinal hernia, flat 96 feet, and crowded toes (Figure 1).

Figure 1 is missing, add a figure 1 or eliminate and change figure 2 into figure 1

**Level of interest**
Please indicate how interesting you found the manuscript:

An article whose findings are important to those with closely related research interests

**Quality of written English**
Please indicate the quality of language in the manuscript:

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