Reviewer's report

Title: Shwachman-Diamond Syndrome: A complex case demonstrating the potential for misdiagnosis as Asphyxiating Thoracic Dystrophy (Jeune Syndrome)

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Reviewer: Marco Cipolli

Reviewer's report:

The article is well written and approaches the important issue of SDS differential diagnosis. The paper is not a novel contribution to existing knowledge on SDS but the case discussed here is a good and clear example of how to improve diagnostic capacity and how to avoid clinical mistakes.

I have some comments for the authors which I have outlined below.

Major Compulsory Revisions

Background

Pg 3. Line 18. As regards the haematological involvement, it is necessary to underline that the most important sign is represented by neutropenia. In fact, this is one of the two main characteristics for the clinical diagnosis and is generally present in over 80% of SDS patients, also at the time of diagnosis.

Case presentation

Pg 5. Were any haematological data available at birth and during the first admission to the neonatal clinic? Is it possible to find out whether neutropenia was present at that time? The same applies for the admissions at 2 and 5 years. Only the platelet count is mentioned in the text.

The authors assert that the patient became pancytopenic at 10 years, but they don’t show any earlier data, except for platelets. This may be confusing, one could extrapolate that the main clinical signs of the disease start later in life, while we really do not have enough information until 10 years of age.

Have the authors any data on foetal haemoglobin?

As for pancreatic function and growth, it is well known that in about 50% of SDS patients, pancreatic activity improves after 5 years of age. No information about pancreatic activity and absorption before diagnosis is provided. It may be useful to include a figure showing the growth charts of the patient, which would also give the reader some indirect information about her absorption capacity.

Conclusions

Line 3. I would be more cautious in the comment “many of the characteristic symptoms….will not be apparent soon after birth”. Frequently the problem is that
signs or symptoms of SDS are not looked for or not taken into consideration enough by physicians. This is probably what happened for the case described and the diagnosis was probably possible at birth or very soon after. It’s also important to underline that as time passes the diagnosis might become more difficult, particularly if the pancreas starts to improve and the haematological complications are not so severe, as in this case.

The authors make a good point when they state there is a risk of continuing to associate PI with ATD, therefore increasing the risk of a misdiagnosis.

Minor Essential Revisions

As regards the figures, it would be better to add some arrows to indicate and better explain what is written in the legends.

**Level of interest:** An article of importance in its field

**Quality of written English:** Acceptable

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**

I declare that I have no competing interests