Reviewer's report

**Title:** Clinico-pathogenetic findings and management of chondrodystrophic myotonica (Schwartz-Jampel syndrome)

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**Reviewer:** Umbertina Reed

**Level of interest:** A paper whose findings are important to those with closely related research interests

**Advice on publication:** Accept after discretionary revisions

a) Discretionary revisions

The recent discovery that Schwartz-Jampel syndrome (SJS) is caused by mutations in the gene encoding perlecan, makes the present report very interesting and recommended for publication, as there are only a few works focusing the molecular diagnosis of the disorder. Although such works have been referred by the authors, it would be useful: 1. to detail a little more the clinical aspects of SJS, particularly the clinical variability 2. to discuss the subdivision in IA and IB, proposed by Giedion et al (1997) based on the age at onset, the degree of bone dysplasia and the clinical severity. Even considering that type II is definitively out of SJS spectrum, therefore invalidating the subdivision in Types 1 and 2, the characterization of A and B phenotypes could be maintained and perhaps could be associated to different mutations at 1p36.1. As among the 11 references mentioned by the authors, all the most recent are related with molecular or pathogenetic findings, I would suggest: 3. a wider clinical revision possibly including some of the reports published in the last years.

In addition, I suggest: 4. to discuss the result of the treatment at the end of Case Presentation; 5. if possible, to describe more details about the molecular findings (methodology and results).

b) Compulsory revisions

None

**Competing interests:**

None declared.