Reviewer's report

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

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Reviewer: Bertrand Fontaine

Level of interest: A paper of considerable general medical or scientific interest

Advice on publication: Accept after discretionary revisions

In their paper, Ho and colleagues describes the case of a child with Schwartz-Jampel syndrome. The diagnosis is appropriate and the case well-described. In addition, they report the molecular analysis of the patient which shows a decreased expression of perlecan on muscle biopsy and heterozygous mutations in the perlecan gene at splice-site junction sites. This findings are novel, original and well-described.

Minor corrections:
1) myotonia instead of myotonica (title and text)
2) please use the chemical names of the medication instead of their brand names, and indicate dosages
3) please indicate in the description of the texte the absence of consanguinity and the clinical examination of the parent

Competing interests:

None declared.