Title: The Dunnigan-type of familial partial lipodystrophy in the adolescent girl - a case report

Version: 2
Date: 18 September 2015

Reviewer: Véronique Béréziat

Reviewer's report:

Krawiec et al reported a case of a 14-year old girl addressed to the Paediatrics Department due to chronic hyper transaminasemia. She had all the physical signs of lipodystrophy, confirmed by clinic examination. Genetic screening has evidence the R482W mutation in the LMNA gene, typically found mutation in 80% of patients.

Major Compulsory Revision

If the case report is well built with a precise description of the clinical data, my main criticism remains that the authors do not put enough evidence in the uniqueness of this case.

As mentioned by the authors in the discussion section, three cases of FPLD2 have already been described in Poland, including a girl of 17 years-old (Ref 1). Moreover, some 85% of FPLD2 patients present an heterozygous missense substitutions at LMNA codon 482 (Ref 2). However, authors should have one or two paragraphs indicating why this case is unique, as required in the CARE Checklist.

Minor Essential Revisions

1. Familial partial lipodystrophy of the Dunnigan type must be refer as FPLD2.
2. Page 3 line 9 the authors indicated that the LMNA gene encodes lamins instead of A-type lamins (lamin A and C). The authors should correct the sentence.
3. Page 3 lines 14-16 “holistic approach to a patient” is written twice, the authors should correct this.
4. Page 3 line 23 fist should be replaced by first.
5. When describing the genetic test performed page 5 the authors should add a figure with the DNA sequencing revealing the heterozygous substitution.
6. In the discussion, the authors indicated that FPLD2 incidence was 1 case per 15 millions. In fact, the incidence of the disease is rather than 1 in 200,000 (Ref 3).
7. Some references are missing.
   • Page 3, lines 8-10, the authors should include a review which address the structural and functional role of lamin A/C instead of a clinical review.
• They also should cite the two original papers about the LMNA p.R482 heterozygous substitutions (Ref 4-5).

References
3- Al-Shali K.Z. and Hegele R.A. , ATVB. 2004 ; 24 : 1591–95

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:
I declare that I have no competing interests.