Reviewer’s report

Title: Gene Expression Analysis Reveals Impaired Mitochondriogenesis and Adipogenesis in Adipose Tissue from a Patient with Acquired Partial Lipodystrophy (Barraquer-Simons Syndrome): a case report

Version: 2 Date: 20 May 2008

Reviewer: Jacqueline Capeau

I am familiar with the literature and believe that this case meets one of the 9 criteria for evaluation in the journal: Findings that shed new light on the possible pathogenesis of a disease or an adverse effect

Has the case been reported coherently?: No

Is the case report authentic?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

> JP Guallar et al have performed an analysis of the mRNA expression
> of a number of genes in an adipose tissue sample from a patient
> with acquired partial lipodystrophy as compared to 10 controls.
> They observe that the expression of some genes is decreased while
> that of others is not modified. They conclude to altered
> adipogenesis and mitochondrial function.
> This study is original and I have not found similar studies in APL
> patients. Therefore, the topic is interesting. However, some
> important biological values are missing to complete the evaluation
> of this patient. In addition, it would be important to provide a
> morphological analysis of the adipose tissue and to complete the
> gene expression study.
>
> Specific points
> It is important to give the results of the patient’s metabolic
> parameters: glycemia, lipids, insulinemia. Even if such alterations
> are uncommon in patients with a Barraquer-Simons syndrome, they are
> present in some of them. In addition, the authors explain that the
> patient has a progeroid appearance which is not common in APL
> patients (but facial lipoatrophy can give an impression of aging)
> and a male aspect. Therefore it is important to search for the
> presence of hyperandrogenism at the clinical and biological level.
> It would be interesting to search for mutations in the lamin genes
> inasmuch as muscular signs were noticed which could be related to
> muscular dystrophy. The patient has hepatic steatosis: liver
> functions can be also presented. A photo would be interesting to
> insert.
>
> Since the expression of adiponectin but not leptin is decreased in
> adipose tissue, the circulating level of the two adipokines have to
> be given.
>
> The patient had an adipose tissue biopsy from the arm. Due to
> generally severe lipoatrophy in the upper part of the body, the
> authors have to confirm that they were able to obtain fat and that
> mRNA was indeed extracted from adipose tissue and not connective
> tissue. It would be important to provide a picture of fat
> morphology to assess whether the size of adipocytes was normal,
> homogeneous, whether fibrosis was present and/or macrophages.
>
> The authors find that PPARgamma was decreased as a transcription
> factor together with decreased expression of PPARgamma regulated
> genes indicating a decreased adipogenesis.
> However, the expression of the two transcription factors involved
> in mitochondriogenesis is not decreased: therefore the authors
could not assert that mitochondriogenesis is impaired; they have to modify the title and summary.

Since the patients presented altered level of complement, it would be interesting to evaluate the mRNA expression of the complement factors secreted by adipocytes.

**Quality of written English:** Needs some language corrections before being published

**Declaration of competing interests:**

I declare that I have no competing interests