

PEER REVIEW HISTORY

BMJ Paediatrics Open publishes all reviews undertaken for accepted manuscripts. Reviewers are asked to complete a checklist review form and are provided with free text boxes to elaborate on their assessment. These free text comments are reproduced below.

This paper was submitted to a another journal from BMJ but declined for publication following peer review. The authors addressed the reviewers' comments and submitted the revised paper to BMJ Paediatrics Open. The paper was subsequently accepted for publication at BMJ Paediatrics Open.

ARTICLE DETAILS

TITLE (PROVISIONAL)	Incidence of infantile Pompe disease in the Maroon population of French Guiana.
AUTHORS	Elenga, Narcisse; Verloes, Alain; Mrsic, Yajaira; Basurko, Célia; Schaub, Roxane; Cuadro, Emma; Kom-Tchameni, Rémi; Carles, Gabriel; Lambert, Véronique; Boukhari, Rachida; Fahrasmene, Aniza; Jolivet, Anne; Nacher, Mathieu; Benoist, JF

VERSION 1 - REVIEW

REVIEWER	Zurynski, Yvonne The University of Sydney, Australia Competing interests: I have no competing interests
REVIEW RETURNED	01-Sep-2017

GENERAL COMMENTS	<p>A very interesting study of Pompe disease in a specific ethnic community in French Guiana. The study has important implications for genetic screening, genetic counselling and early treatment intervention.</p> <p>The following points need to be addressed:</p> <p>The terms incidence and prevalence seem to be used inconsistently in parts - please ensure that you refer to incidence (or birth prevalence) when referring to the 19 infants and to prevalence when referring to the screened pregnant women.</p> <p>The "usual" prevalence of IPD is given as 1/150,000 - please provide a source or reference for this figure.</p> <p>The annual incidence for French Guiana is skewed because all infants were from Saint-Laurent du Maroni Maternity unit and all had Maroon parents. Are there any data on the incidence of IPD in the non-Maroon community of French Guinea to provide a comparison?</p> <p>The results state that molecular data was available for "several children born after 2010" - how many? be specific.</p> <p>What is meant by "oral information was accompanied by written information? What information was collected from the women and how?</p> <p>It would be worth commenting about the trends over time of IPD and treatment with ERT. Since when has ERT been routinely offered to all IPD infants?</p> <p>These results are clearly important for the Maroon community in French Guiana. Please clarify whether population-based screening would be warranted in other countries of the world and whether this would be recommended only for people of Maroon descent or wider screening?</p> <p>Please edit the manuscript for grammar and English expression e.g.</p>
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	consanguineous "mating" is not acceptable language - "union" would be better. The term "maternities" is a little odd - maternity units or maternity hospitals might be better
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REVIEWER	Mercer, Raul FLACSO (Latin American School of Social Sciences). Program of Social Sciences and Health Buenos Aires, Argentina Competing interests: No competing interests
REVIEW RETURNED	24-Sep-2017

GENERAL COMMENTS	<p>The article is well written. It addresses the problem from the local context and compares the epidemiological information on Pompe disease with data from other countries.</p> <p>For its part, the title of the work refers to "prevalence" while throughout the manuscript refers to "incidence." This aspect has to be clarified for the purpose of integrating the historical component (records) and the prospective (based on the screening study).</p> <p>There is no mention of the screening study in the abstract. In this section, it is suggested that "Methods" should replace "Patients".</p> <p>In the conclusions, reference is made to the value of identifying couples at risk but does not clarify why. (i.e. genetic counseling). This needs to be clarified.</p> <p>The authors refer to the therapeutic approach through enzymatic replacement and, on the other hand, they emphasize the need to implement a screening program. I believe that the position regarding the approach to the problem (treatment vs. prevention) should be clearly stated, at least, in the discussion.</p> <p>The work requires minimal changes.</p>
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VERSION 1 – AUTHOR RESPONSE

Reviewer: 1

A very interesting study of Pompe disease in a specific ethnic community in French Guiana. The study has important implications for genetic screening, genetic counselling and early treatment intervention.

Thank you so much

The following points need to be addressed:

The terms incidence and prevalence seem to be used inconsistently in parts - please ensure that you refer to incidence (or birth prevalence) when referring to the 19 infants and to prevalence when referring to the screened pregnant women.

We totally agree with the reviewer. OK done

The "usual" prevalence of IPD is given as 1/150,000 - please provide a source or reference for this figure.

ok

The annual incidence for French Guiana is skewed because all infants were from Saint-Laurent du Maroni Maternity unit and all had Maroon parents. Are there any data on the incidence of IPD in the non-Maroon community of French Guiana to provide a comparison?

Until now, we found no cases of IPD in the non-Maroon community of French Guiana

The results state that molecular data was available for "several children born after 2010" - how many? be specific.

eight children

What is meant by "oral information was accompanied by written information? What information was collected from the women and how?

What is meant by "oral information was accompanied by a written consent form? What information was collected from the women and how?

Information and consent form.

Before inclusion in the study, information was given to women by the investigators (reading, explanation and delivery of the information note). The participant was free to ask questions about the research and was informed of her right to refuse to participate in the research. The investigator recorded in the patient's medical record that she was informed and consented to be included in the research protocol. This form included the explanation of Pompe disease, the purpose of the study and the blood sampling that will be performed for women.

It would be worth commenting about the trends over time of IPD and treatment with ERT. Since when has ERT been routinely offered to all IPD infants?

OK done

These results are clearly important for the Maroon community in French Guiana. Please clarify whether population-based screening would be warranted in other countries of the world and whether this would be recommended only for people of Maroon descent or wider screening?

Such screening is already done in Taiwan and in some US states. Another approach would be to implement neonatal screening, which currently exists in Taiwan and is announced in some US states.

Please edit the manuscript for grammar and English expression e.g. consanguineous "mating" is not acceptable language - "union" would be better. The term "maternities" is a little odd - maternity units or maternity hospitals might be better

OK done

Reviewer: 2

The article is well written. It addresses the problem from the local context and compares the epidemiological information on Pompe disease with data from other countries.

For its part, the title of the work refers to "prevalence" while throughout the manuscript refers to "incidence." This aspect has to be clarified for the purpose of integrating the historical component (records) and the prospective (based on the screening study).

OK done

There is no mention of the screening study in the abstract. In this section, it is suggested that "Methods" should replace "Patients".

OK done

In the conclusions, reference is made to the value of identifying couples at risk but does not clarify why. (i.e. genetic counseling). This needs to be clarified.

Implementation of a systematic neonatal screening in French Guiana would identify a number of couples at risk and could allow delivering appropriate genetic counselling and prenatal diagnosis.

The authors refer to the therapeutic approach through enzymatic replacement and, on the other hand, they emphasize the need to implement a screening program. I believe that the position regarding the approach to the problem (treatment vs. prevention) should be clearly stated, at least, in the discussion.

Ok done

The work requires minimal changes.