Reviewer’s report

Title: Chart Validation of an Algorithm for Identifying Hereditary Progressive Muscular Dystrophy in Healthcare Claims

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Reviewer: Kan Hor

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Chart Validation of an Algorithm for Identifying Hereditary Progressive Muscular Dystrophy in Healthcare Claims

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Reviewer Comments:
General comments: In their manuscript entitled "Chart Validation of an Algorithm for Identifying Hereditary Progressive Muscular Dystrophy in Healthcare Claims" the authors attempted to use a coding algorithm from a large commercial claims database to identify patients with hereditary MD and attempt to characterize patients with MD. The idea behind this is to better understand the hereditary MD population in order to understand the treatment algorithm. I agree with the authors that this is an important process as hereditary MD do not yet have specific International Classification of Disease-9/-10 codes. The overall conclusion is that the "case-finding algorithm accurately identified patients with MD, primarily Duchenne MD, within a large administrative database. Overall the study designed is well thought out and I agree with the authors that in the rare disease space understanding the existing patient population is important especially in informing disease natural history and ensuring the right population is chosen for trials (ie age and disease status and comorbidities).
I agree with the author's limitation section.

This is a rather straightforward descriptive study that serve the purpose of the hypothesis. Overall I think this is an important paper to publish as a pathway to better understand (1) how challenging the coding system is to identify disease (2) the importance to understand rare disease through large database to inform how to best choose the right population for trials. This paper in many way is similar to a recently published paper by Soslow et al, 2019 in Cardiology in the Young.
There are some issues I would like the authors' to address:

Specific Comments:
1. I would however be cautious with how the authors define disease prevalence in the different areas and in particular cardiac disease prevalence. I do not disagree how the author words it but would recommend that there should be some comments that depends on how one defines the specific disease (ie cardiac, respiratory, bone-health and endocrine-related conditions). For example for cardiac disease prevalence if you use ECG you would have a high rate of disease prevalence based on relative
tachycardia and other non-specific ECG findings or if you use systolic function by shortening fraction you would not demonstrate cardiac disease until later. The first shows a high prevalence of disease early using a very sensitive test such ECG but loses specificity. On the other hand using ECHO shortening fraction which is first a blunt tool and second global function decline by shortening fraction occurs later and you underestimate prevalence. I would ask that the authors in the limitations at least address the tools to define disease may alter prevalence and is important in understanding the tool one uses clinically as well as in trials.

2. Similar to (1) above can the authors further detail how a patient is given a disease (ie cardiac, respiratory, bone-health or endocrine issues)? Is it based on ICD coding? It would again be important to address some limitations for how each patient is given a disease. If the authors used specific disease code, medication, specific health care physician who saw the patient.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

I am able to assess the statistics

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