Reviewer's report

Title: Genetic diagnosis of X-linked dominant hypophosphatemic rickets in a cohort study: Tubular reabsorption of phosphate and 1,25(OH)2D serum levels are associated with PHEX mutation type

Version: 3 Date: 28 June 2011

Reviewer: Yves Sabbagh

Reviewer's report:

The authors provide a report regarding 36 index cases with hypophosphatemic rickets. The authors have identified 36 mutations in a very clear manner with emerging techniques that have helped identify mutations that would have been missed using classical methods of mutation detection, namely regular sequencing. Another important point that the paper makes is the presence of hyperparathyroidism in XLH patients prior to the start of treatment. The authors also demonstrate genotype-phenotype correlations between type of mutations with TRP and 1,25(OH)2D3 serum levels. This is the first report to demonstrate 100% of mutation identification in the PHEX gene in patients diagnosed with hypophosphatemic rickets.

The authors have addressed the major issues of the paper and is now acceptable for publication.

Minor Essential Revisions

In discussion paragraph starting with "Hyperparathyroidism is a rather..."
XLHR is mispelled twice as HXLR in the 4th and 5th sentence.
Also hiperoxaliuria should be hyperoxaliuria

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

'I declare that I have no competing interests'