

## **Reviewer's report**

**Title:** A novel HSF4 gene mutation (p.R405X) causing autosomal recessive congenital cataracts in a large consanguineous Pakistani family

**Version:** 1 **Date:** 26 June 2008

**Reviewer:** James Hejtmancik

### **Reviewer's report:**

This is an interesting manuscript in which the authors identify a novel mutation in HSF4 that is associated with autosomal recessive cataracts in a Pakistani family. The question and limitations of the study are well defined, the methods are appropriate and well described, the data are sound, and there are no problems with reporting and deposition of data. The authors provide concise and useful summaries of previous work in this area and the title and abstract accurately convey the essence of the report. There are a number of English grammatical and usage problems in the manuscript. It would benefit from close reading by a line editor, but these are relatively minor in nature. A few specific comments follow. They should be considered minor essential revisions:

1. **Methods, Linkage analysis:** The marker allele frequencies and their source should be given.
2. **Results:** Were any unrelated controls sequenced? While this is less important for a stop mutation in a gene known to cause cataracts, it should still be done.

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

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

**Statistical review:** Yes, and I have assessed the statistics in my report.

**Declaration of competing interests:**

'I declare that I have no competing interests'