Reviewer’s report

Title: Application of targeted multi-gene panel testing for the diagnosis of inherited peripheral neuropathy provides a high diagnostic yield with unexpected phenotype-genotype variability

Version: 2 Date: 30 June 2015

Reviewer: Heather McLaughlin

Reviewer’s report:

Antoniadi and colleagues describe the implementation of a 56 gene next-generation sequencing (NGS) panel for inherited peripheral neuropathy (IPN) in 448 patients, the majority of whom met UKGTN approval criteria. This is a nicely written manuscript describing the diagnostic utility of expanded NGS-based panels for IPN, a spectrum of diseases known for their clinical and genetic heterogeneity. Following the changes implemented by the authors based upon my initial review, I believe this manuscript is suitable for publication in BMC Medical Genetics.

Major Compulsory Revisions
None to report

Minor Essential Revisions
None to report

Discretionary Revisions
None to report

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 am an Senior Clinical Scientist at GeneDx, a fee-for-service clinical genetic testing laboratory.