

ORAL PRESENTATION

Open Access

# Whole-genome sequencing and disease-gene detection

Lynn B Jorde

From *Beyond the Genome 2012*

Boston, MA, USA. 27-29 September 2012

Whole-genome sequencing (WGS) offers unique opportunities to identify rare variants that cause disease. We have developed a new software tool, VAAST (Variant Annotation, Analysis and Search Tool) that permits the identification of specific disease-causing mutations in WGS data. VAAST unambiguously identifies two disease-causing mutations in a family quartet in which both offspring have autosomal recessive primary ciliary dyskinesia and Miller syndrome. In addition, VAAST has identified a new X-linked progeria-like syndrome (Ogden syndrome) using exome data from two unrelated families. The mutation occurs in *NAA10*, which encodes an N-acetyltransferase needed for N-terminal acetylation of proteins. Functional studies demonstrate that the mutation causes a loss of function, and a genetic test has been developed for Ogden syndrome. We have also used VAAST to identify *GATA4* as the cause of cardiac septal defects in a single four-generation pedigree. Using the Utah Population Database, we have identified a large multigenerational pedigree in which VAAST, combined with analysis of shared genome segments, identifies a new locus for Crohn disease. Finally, we present an application of VAAST in the identification of *ATPIA3* as a causal gene for alternating hemiplegia of childhood.

Published: 1 October 2012

doi:10.1186/1753-6561-6-S6-O7

Cite this article as: Jorde: Whole-genome sequencing and disease-gene detection. *BMC Proceedings* 2012, **6**(Suppl 6):O7.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at  
[www.biomedcentral.com/submit](http://www.biomedcentral.com/submit)



Department of Human Genetics, University of Utah School of Medicine, UT, USA



© 2012 Jorde; licensee BioMed Central Ltd. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/2.0>), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.