An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge - DTU Orbit (10/12/2018)

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**Background:** There is tremendous potential for genome sequencing to improve clinical diagnosis and care once it becomes routinely accessible, but this will require formalizing research methods into clinical best practices in the areas of sequence data generation, analysis, interpretation and reporting. The CLARITY Challenge was designed to spur convergence in methods for diagnosing genetic disease starting from clinical case history and genome sequencing data. DNA samples were obtained from three families with heritable genetic disorders and genomic sequence data were donated by sequencing platform vendors. The challenge was to analyze and interpret these data with the goals of identifying disease-causing variants and reporting the findings in a clinically useful format. Participating contestant groups were solicited broadly, and an independent panel of judges evaluated their performance.

**Results:** A total of 30 international groups were engaged. The entries reveal a general convergence of practices on most elements of the analysis and interpretation process. However, even given this commonality of approach, only two groups identified the consensus candidate variants in all disease cases, demonstrating a need for consistent fine-tuning of the generally accepted methods. There was greater diversity of the final clinical report content and in the patient consenting process, demonstrating that these areas require additional exploration and standardization.

**Conclusions:** The CLARITY Challenge provides a comprehensive assessment of current practices for using genome sequencing to diagnose and report genetic diseases. There is remarkable convergence in bioinformatic techniques, but medical interpretation and reporting are areas that require further development by many groups.

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