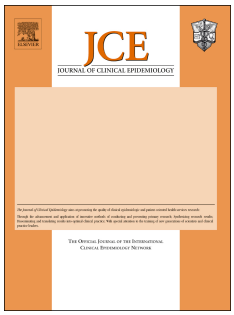
Accepted Manuscript



Controversy and Debate on Clinical Genomics Sequencing - Paper 2: Clinical Genome-Wide Sequencing: Don't Throw Out the Baby With The Bathwater!

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PII: S0895-4356(17)31010-7

DOI: 10.1016/j.jclinepi.2017.08.020

Reference: JCE 9473

To appear in: *Journal of Clinical Epidemiology*

Received Date: 12 January 2016

Revised Date: 30 August 2016

Accepted Date: 24 August 2017

Please cite this article as: Adam S, Friedman JM, Controversy and Debate on Clinical Genomics Sequencing - Paper 2: Clinical Genome-Wide Sequencing: Don't Throw Out the Baby With The Bathwater!, *Journal of Clinical Epidemiology* (2017), doi: 10.1016/j.jclinepi.2017.08.020.

Clinical Genome-Wide Sequencing:

Don't Throw Out the Baby With The Bathwater!

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In patients and families with serious diseases produced by *de novo* or inherited mutations, optimal clinical care requires characterization of the underlying genetic pathology. Without a precise diagnosis, affected individuals and their families do not know whether the problems will resolve or grow worse with time, or what additional problems may arise in the future. No treatment, or only limited symptomatic therapy, is usually available without a precise diagnosis, and the genetic counselling and prenatal diagnosis available to such families are usually very general and unsatisfactory.

Clinical management of patients with suspected genetic disease therefore begins with complete description of the clinical features and a search for the precise genetic cause. Occasionally, specific features in a patient are characteristic of a particular genetic cause that accounts for almost every case. This occurs with cystic fibrosis, for example. In such instances, a locus-specific genetic test can be done to confirm the diagnosis. Much more often, however, the condition may have many