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A new era in the interpretation of human genomic variation

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In a commentary published today in *Genetics in Medicine*, Heidi Rehm, PhD, highlights the pressing need for standardized human genomic variant interpretation and calls on more stakeholders to join the data sharing movement.

"It is time we considered the sharing of variant interpretations a fundamental right of patients to ensure that they receive accurate clinical care," writes Rehm. "We must ensure that the professional opinion of geneticists is based on a fully shared body of evidence with open andongoing community review, and that we support this endeavor in a cost-effective manner."

Rehm is the principal investigator of ClinGen - the Clinical Genome Resource - which has launched a public list of genetic testing labs that meet minimum data sharing requirements this week. Payers are beginning to require data sharing as a condition of reimbursement - some providers are ordering tests only from labs that are on the ClinGen list.

In her commentary, Rehm discusses how standards and strategies for variant interpretation have evolved dramatically over the past five years. The commentary also highlights four GIM articles describing consensus building projects that have improved consistency in variant interpretation (Harrison et al., Lebo et al.) as well as analyses and resources to further support variant interpretation (Nykamp et al, Yang et al).

Rehm concludes her commentary with a call to action - to regulatory agencies, payers, healthcare providers, journals and clinical laboratories - to require data sharing.

Payers are beginning to require data sharing as a condition of reimbursement. The genetics community has demonstrated that data sharing improves genetic interpretation. Some providers have already decided to only order testing from labs that share data. Aetna has passed a policy to only reimburse breast cancer genetic tests from labs that share data and others are following suit.

More information: Heidi L Rehm, A new era in the interpretation of human genomic variation, *Genetics in Medicine* (2017). DOI: 10.1038/gim.2017.90

Nykamp K, Anderson M, Powers M, et al. Sherloc: a comprehensive refinement of the ACMG-AMP variant classification criteria. *Genet Med* 2017

Harrison SM, Dolinsky JS, Knight Johnson AE, et al. Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. *Genet Med* 2017

Lebo MS, Zakoor K-R, Chun K, et al. Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant interpretation comparisons through the Canadian Open Genetics Repository (COGR). *Genet Med* 2017

Yang S, Lincoln SE, Kobayashi Y, et al. Sources of discordance among germ-line variant classifications in ClinVar. *Genet Med* 2017

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