Patient re-contact after revision of genomic test results: A new ACMG points to consider

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Genomic testing is becoming increasingly common in medicine. Moreover, ongoing advances in technology and an ever-increasing understanding of what genetic variants mean can result in reinterpretation of the clinical significance of variants found in patients. This can occur when the patient's indications for the original test are unchanged or when their phentoypes or family histories require a broader reanalysis or repeat of the test.

What should be done when there is a discovery of a new and important relationship between a disease and a genetic variant in a patient who has previously undergone genetic/genomic testing? This complex question creates uncertainty for the ordering physician, the clinical laboratory and the patient. No definitive answers currently exist, but legal, ethical and practical issues need to be considered.

The ACMG's new "Patient re-contact after revision of genomic test results: points to consider—a statement of the American College of Medical Genetics and Genomics (ACMG)" is intended to help providers to develop policies/procedures regarding re-contact appropriate to individual practice settings and applicable to each patient/family circumstance.

It states in part, "Changes in interpretation of complex clinical genomic test results are inevitable. Ultimately, the ordering health care provider, clinical geneticist, clinical laboratory, referring specialty and primary care physician, patient and family may each have a role regarding re-

contact. These expectations should be explicitly delineated as part of the informed consent process before the sample is obtained and reviewed again when disclosing initial results."

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