# ACMG Secondary Findings: A Reporting Dilemma

BAYLOR GENETICS

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## OBJECTIVES

Since 2013, the American College of Medical Genetics and Genomics (ACMG) has provided guidance for laboratories regarding the reporting of specific clinically actionable secondary findings. These recommendations apply to patients who undergo sequencing tests such as whole exome sequencing or whole genome sequencing (WES/WGS). To date, ACMG has not provided specific guidance as to how these findings should be reported in the context of duo or trio WES/WGS for which parental secondary finding results need to be considered. There have been anecdotal reports of family members encountering difficulties with receiving treatment or treatment coverage because their results are within a proband's report. In addition, including all member's results in one report may conflict with HIPAA privacy protections as medical records will include PHI from other members. Here, a review of laboratories was performed to identify how these entities handle secondary findings and options for reporting that are available to providers and patients using these laboratories.

## METHODS

A review of the Genetic Testing Registry (GTR) was performed in April 2023 using the terms "exome" or "genome" with "Laboratories" selected and Lab location set to "United States". The website for each laboratory was reviewed to confirm that they performed clinical duo or trio WES/WGS. Information regarding mechanism for patient consent to report secondary findings, whether individual reports were issued for proband and family members, if separate (independent) reporting of secondary findings was available for proband and family members, and if there were additional costs for independent reports. Where necessary, laboratories were contacted directly by email or phone call to obtain or clarify information. All commercial laboratories where sufficient information was obtained were included. Several academic laboratories were also selected to be included in this review based on available information. The data from each of these labs were anonymized. The costs for reports were rounded.

## TABLE 1

| | | | | |
|---|---|---|---|---|
| Baylor Genetics | GeneDx | Perkin Elmer | Prevention Genetics | Variantyx |
| Rady Children's | ARUP | LabCorp | Blueprint | Quest |
| Mayo Clinic | Centogene | Ambry | Knight Diagnostics | Greenwood |

**Table 1:** The labs that were included in this review on secondary finding practices.

## FIGURE 1

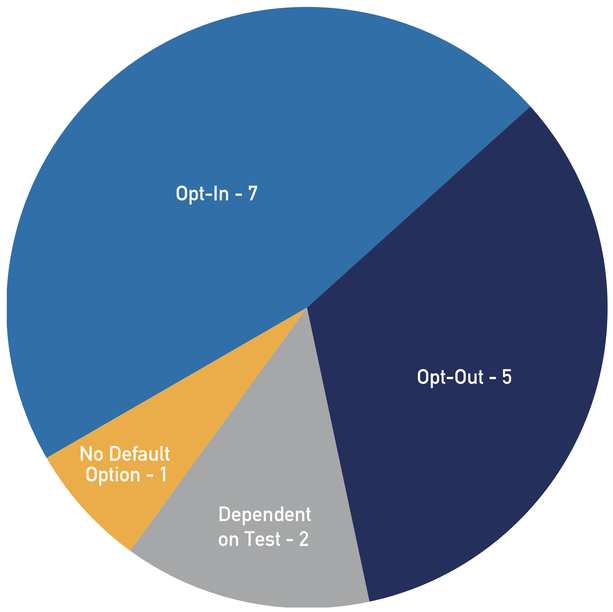


**Figure 1:** Breakdown of how each laboratory reviewed allows for patients to consent into reporting of secondary findings.

## FIGURE 2

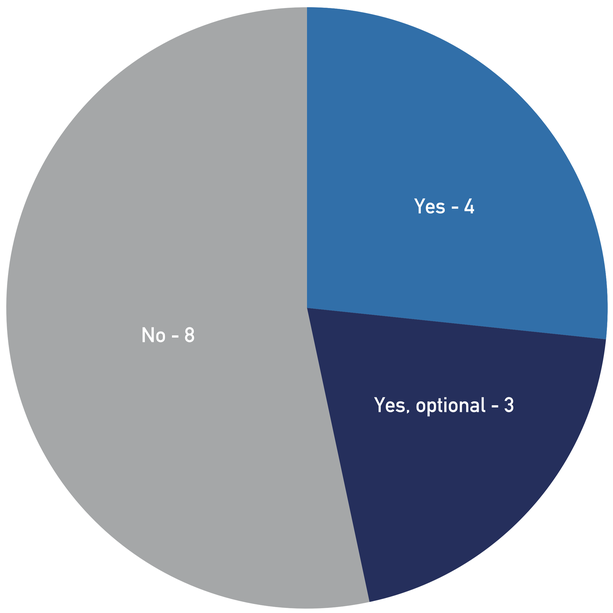


**Figure 2:** Breakdown of whether each laboratory reviewed allows for individual reports to be issued for the proband as well as their family members.

## FIGURE 3

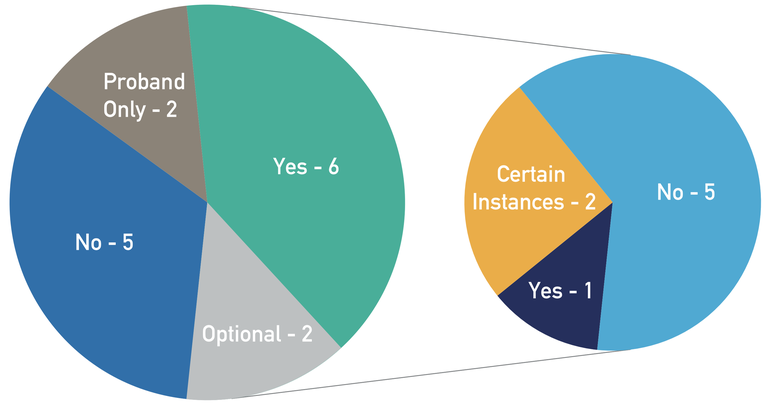


**Figure 3:** Breakdown of whether each laboratory performs independent secondary finding analysis (left pie). For those that do or offer the option to do so, whether this is charged is shown (right pie).

## RESULTS

Data from 15 laboratories were obtained (Table 1). Among the reviewed laboratories, consenting, reporting, and billing practices vary. 12/15 utilize either a default opt-in or opt-out approach to report secondary findings if the patient does not choose whether these should be reported. However, two determine if secondary findings should be reported based on the WES/WGS test code alone and one has no default option (Figure 1). 8/15 do not issue individual reports with duo/trio tests for probands and family comparators (Figure 2). 8/15 allow for proband and comparators to have independent secondary finding analysis performed, of which 6 do so by default. Of those that allow for independent secondary finding analysis, 3/8 charge at least in some instances for independent reports (Figure 3). Two charge approximately $500 for these reports, the third could not provide an estimate without establishing a client account.

## CONCLUSIONS

As WES and WGS become more commonly utilized, additional studies to further explore how processes surrounding secondary findings are handled by laboratories should be pursued. Given how these findings are reported and billed in certain instances and the potential issues that might arise from doing so, it may be helpful to routinely provide individual reports for ACMG secondary findings to avoid potential HIPAA violations or from impeding the ability for payer coverage of treatment. Providing these reports at no-charge would remove barriers to equitable patient care. To best accommodate the needs of families, the benefits of allowing each family member to independently opt for reporting of secondary findings should be further explored as well.