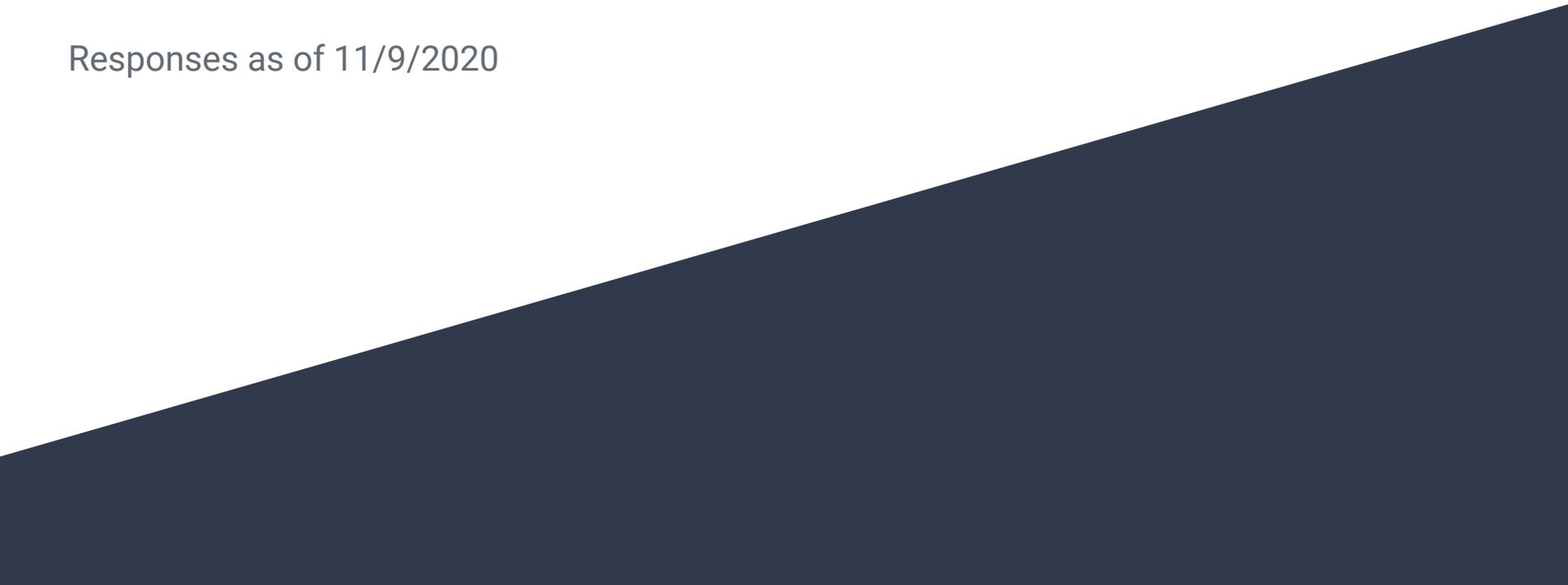


CNV Technical Standards Pre-Webinar Survey Results

Responses as of 11/9/2020

A dark blue diagonal graphic that starts from the bottom left corner and extends towards the top right corner, covering the lower half of the slide.

Attendee Demographics

484 surveys submitted

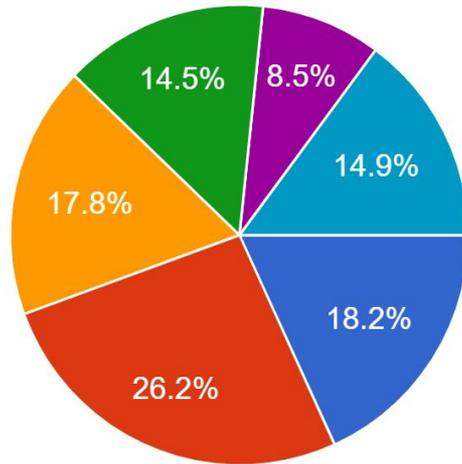
31 Countries attending

- United States
- Canada
- United Kingdom
- Israel
- India
- Australia
- China
- Germany
- Spain
- Norway
- France
- South Korea
- Vietnam
- Iran
- Brazil
- Slovenia
- Scotland
- Qatar
- Mexico
- Oman
- Estonia
- Italy
- Switzerland
- Ireland
- Ukraine
- South Africa
- Nigeria
- Portugal
- Philippines
- Algeria
- Turkey

Demographics

Years of Experience

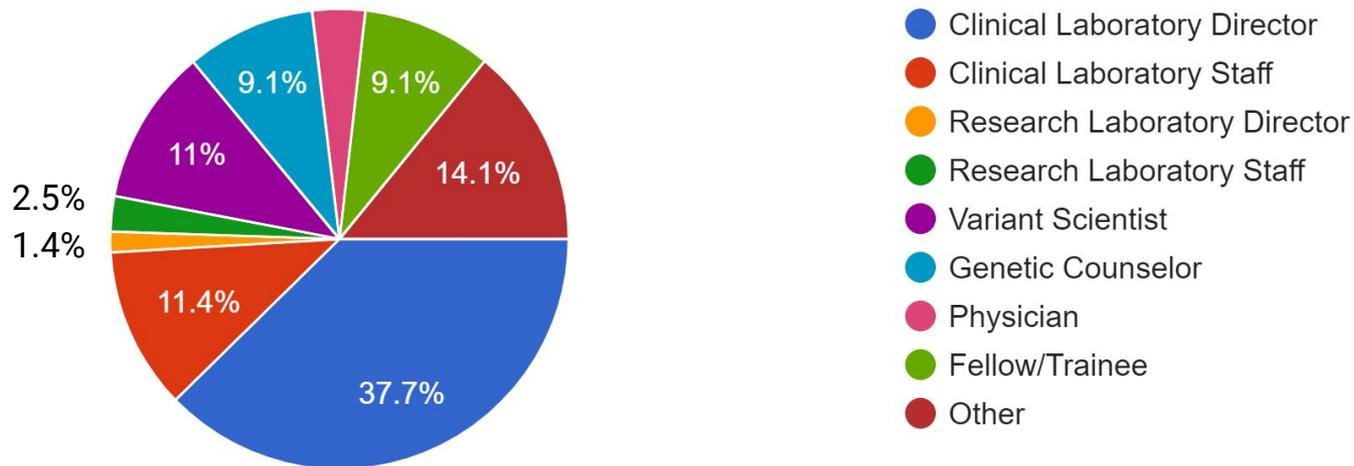
484 responses



Demographics

Please indicate your primary role.

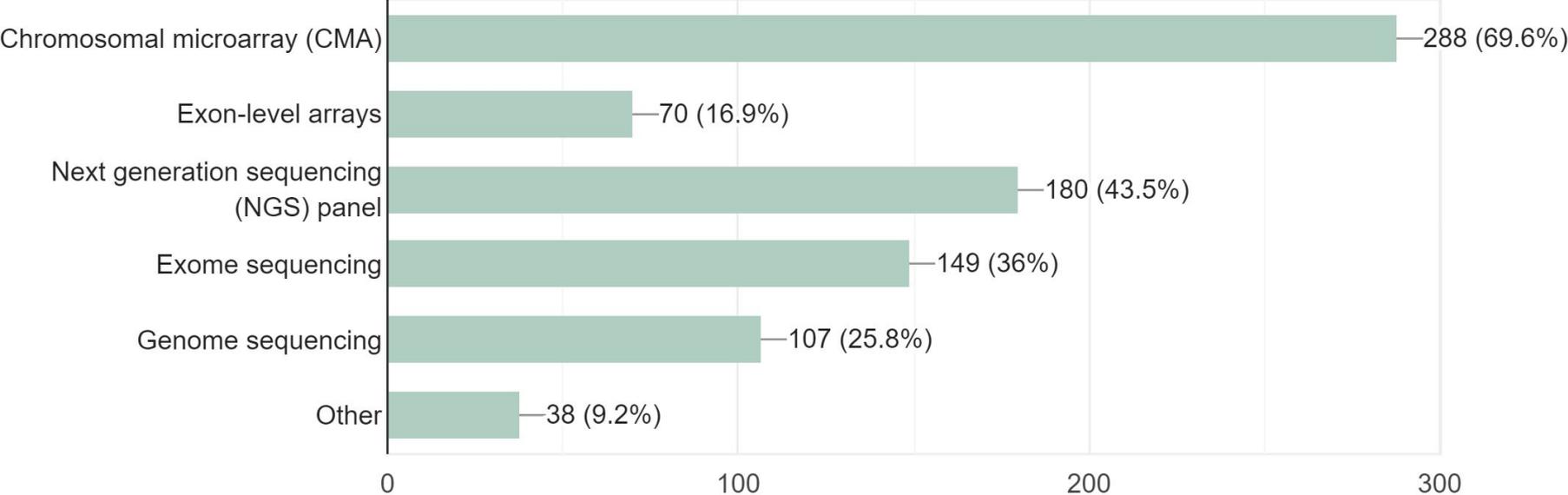
483 responses



Which of the following technologies best represents the scope of your routine CNV workup?

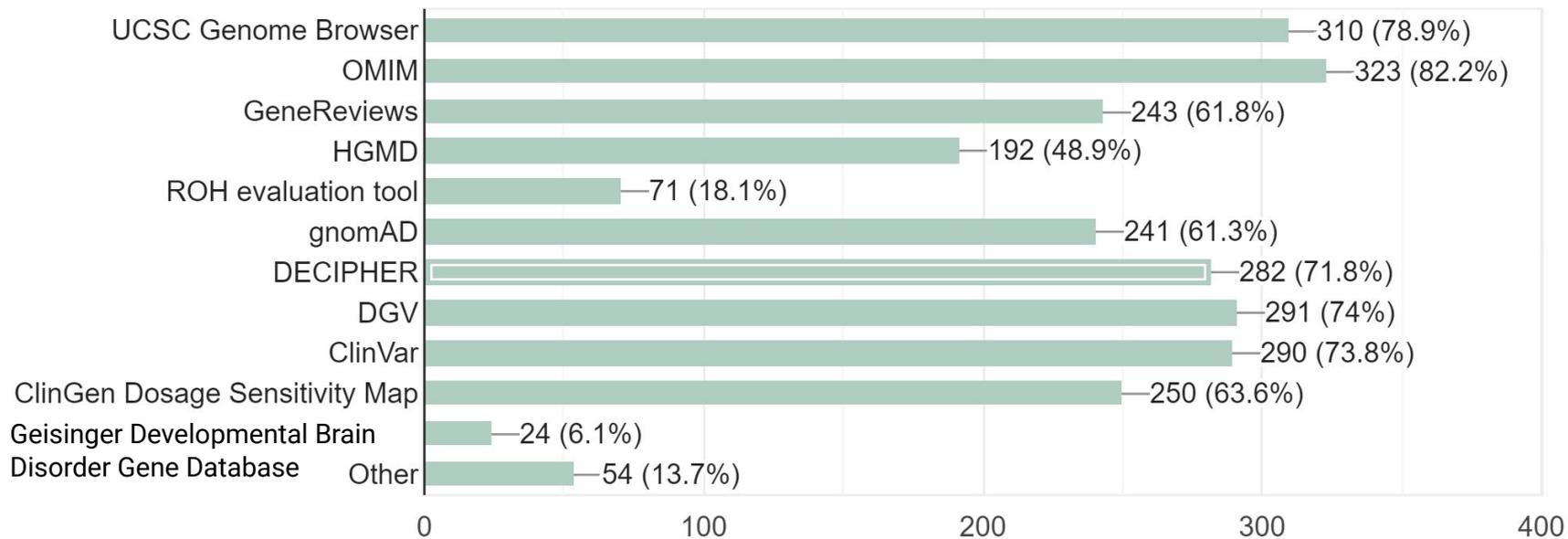
Check all that apply.

414 responses



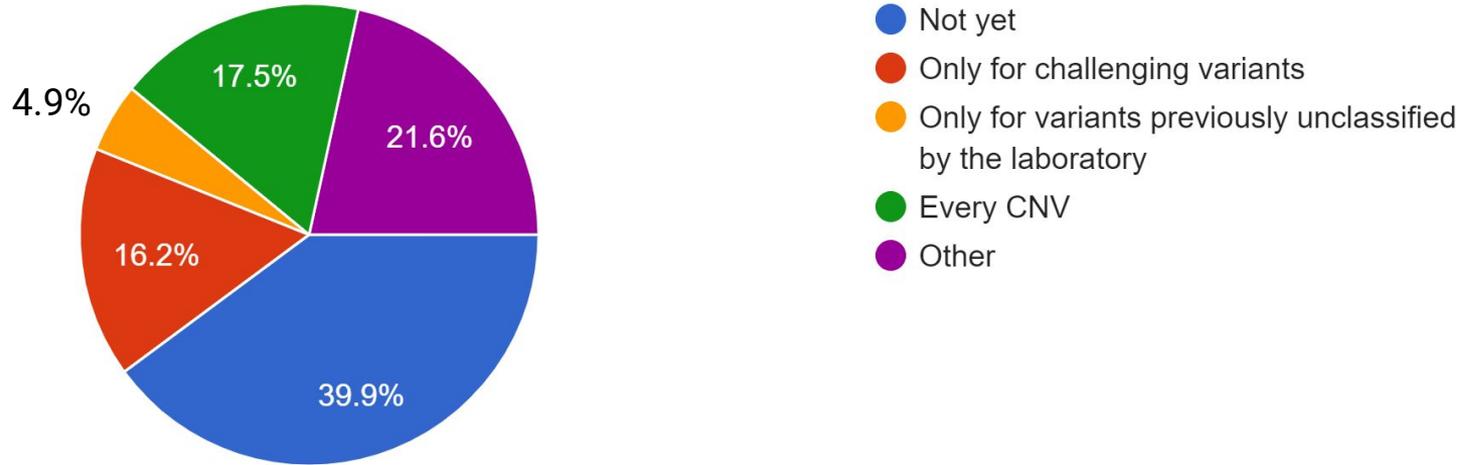
What databases do you use to help with your CNV evaluation? Select all that apply

393 responses



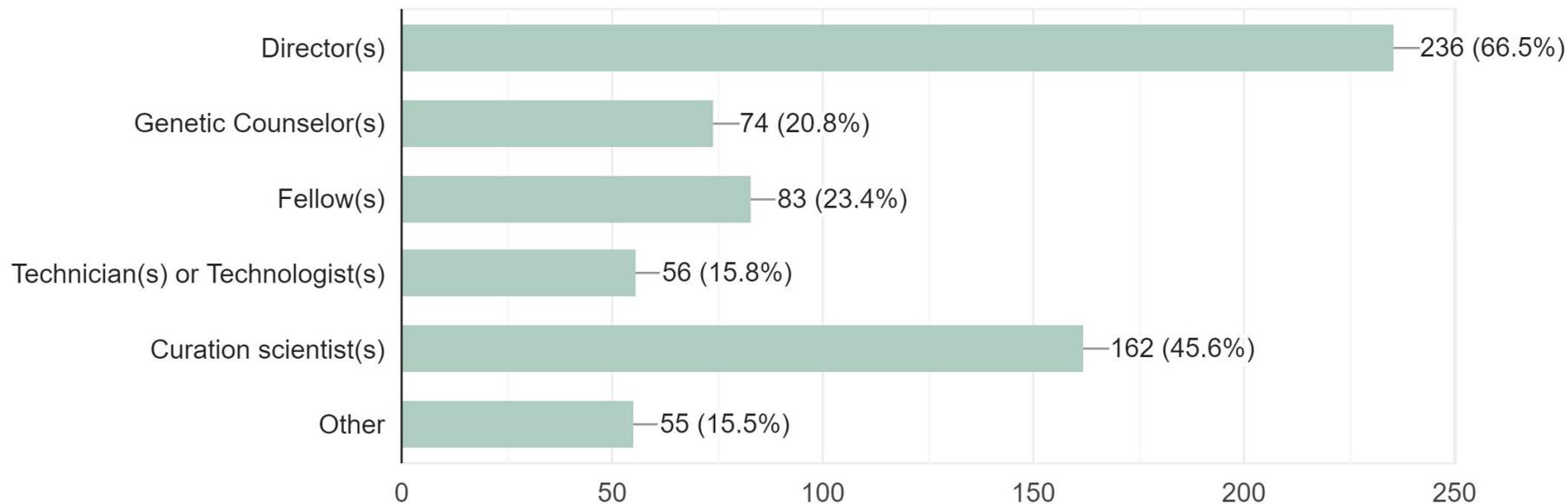
To what level has your laboratory implemented the new guidelines?

371 responses



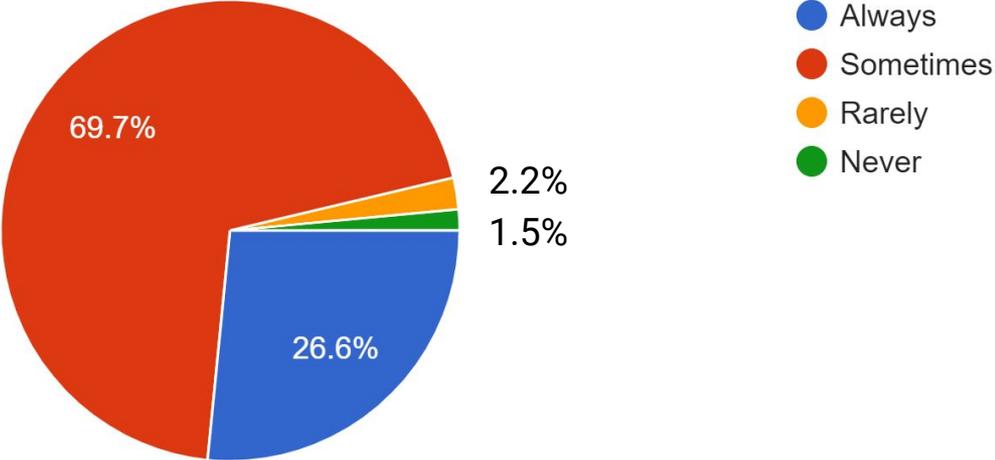
In general, who in the laboratory utilizes the scoring metrics? Select all that apply

355 responses



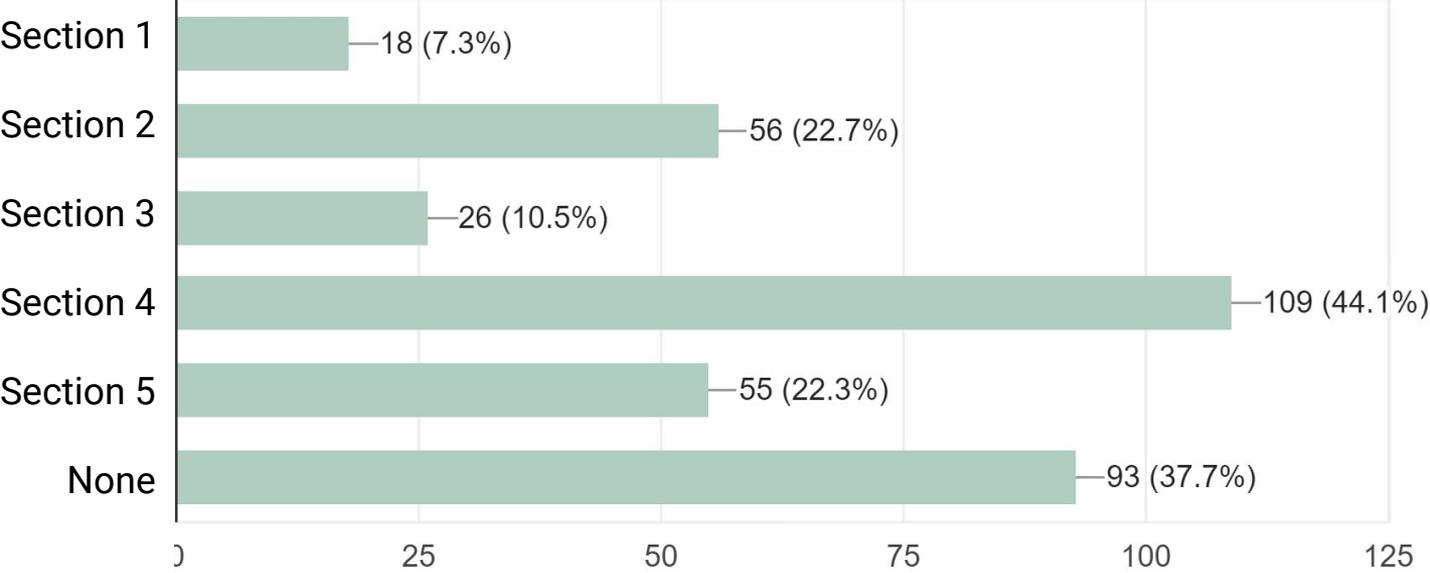
For CNVs where you have used the metric to come to a classification, how often do you agree with the classification?

271 responses



Are there any sections of the metric that have been problematic? Select all that apply.

247 responses



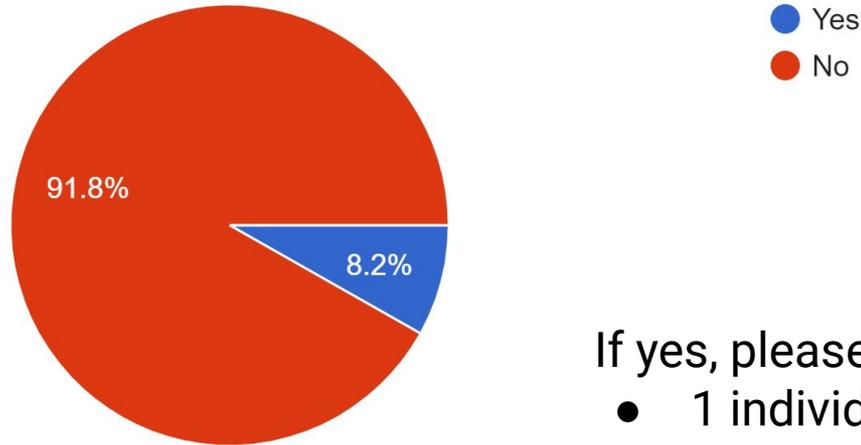
Are there any sections of the metric that have been problematic?

Free text responses

- 4 individuals reported that section 4 can be time consuming and subjective
- 11 individuals stated relevant literature can be tough to obtain and apply to the criteria
- 3 individuals requested the best resources to use for curation
- 5 individuals reported increased numbers of VUS classifications and difficulty getting genes to other classifications
- 1 individual asked which guidelines to use for CNVs with low penetrance
- 3 individuals reported that they had difficulty with X chromosome CNVs

Have the new guidelines changed your practice around requesting parental testing?

245 responses

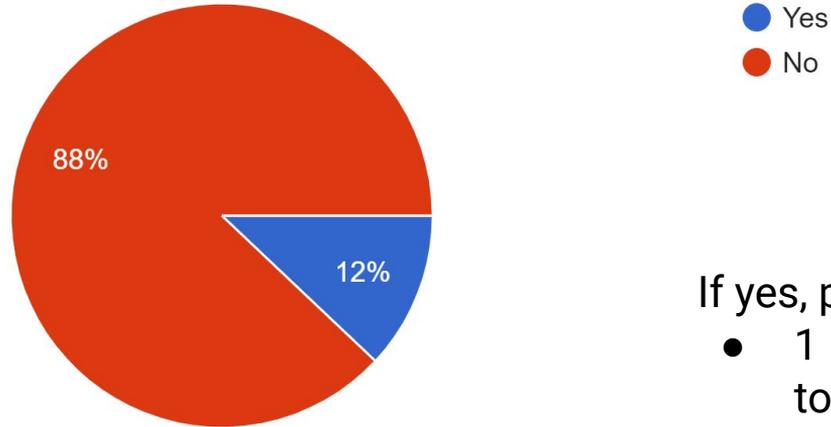


If yes, please describe:

- 1 individual stated they were more likely to request parental testing to answer section 5 of the criteria
- 2 individuals reported requesting parental testing for VUS, LP, and P CNVs

Have you modified the current metric in any way in routine work?

258 responses

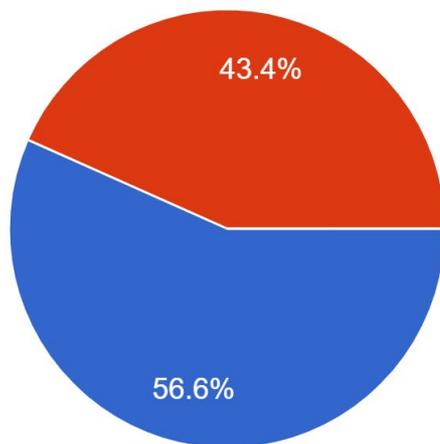


If yes, please describe:

- 1 individual does not apply metrics to small CNVs previously classified as benign
- 2 individuals assign 0 points for number of genes
- 1 individual uses personal judgement to help with classification

Does your lab currently use CNV size criteria to guide reporting practices?

279 responses

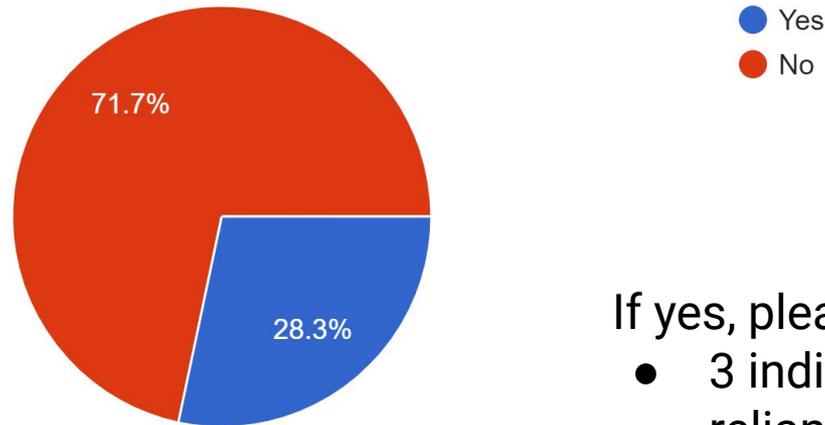


If yes, please describe:

- Responses varied. Some laboratories also indicated different size thresholds for prenatal vs. postnatal.
 - Prenatal:
 - Deletions: 200 kb to >1Mb
 - Duplications: 500 kb to >2Mb
 - Postnatal:
 - Deletions: >25kb to 250kb
 - Duplications: >50kb to 500kb

Has or will this process (of using CNV size criteria to guide reporting practices) change based on the new guidelines?

233 responses



If yes, please describe:

- 3 individuals stated they were less reliant on size criteria
- Reporting differently: one individual is reporting slightly fewer VUS, and one is reporting fewer LP.

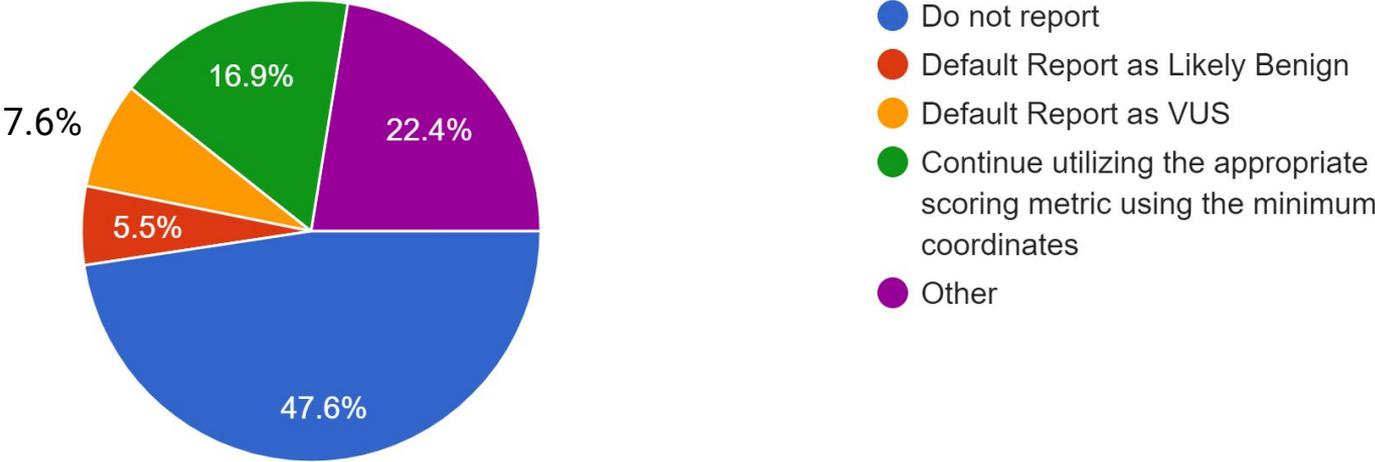
Using general population data (e.g. DGV or gnomAD SV), what's your labs established cutoffs for scoring a CNV as benign?

237 responses



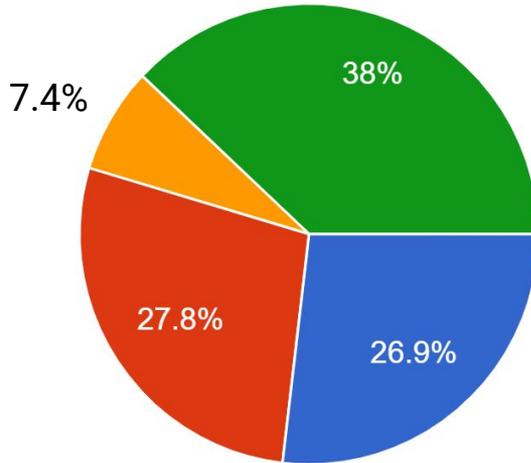
How does your lab approach CNVs over non-protein coding genes/intronic regions?

254 responses



If your method of detection is CMA, how does your lab handle intragenic or partial duplications of established HI genes?

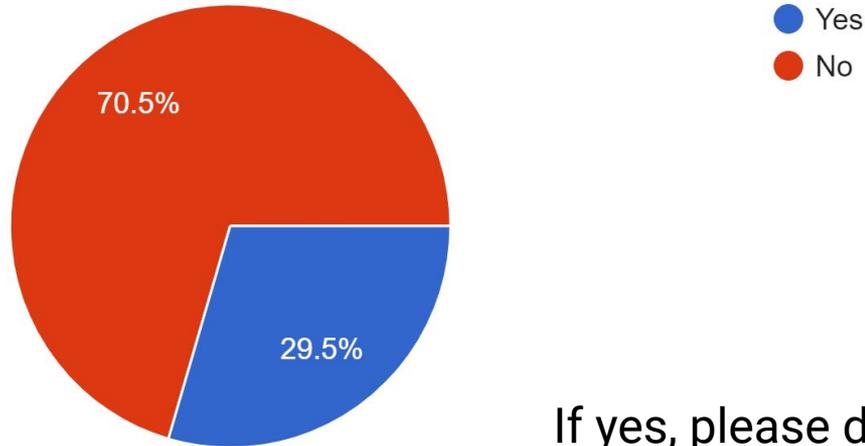
216 responses



- Default to VUS- if exact breakpoints are unknown, we don't feel comfortable utilizing the metric further
- Continue utilizing the DUPLICATION scoring metric using the minimum coordinates
- Continue utilizing the DELETION scoring metric using the minimum coordinates
- Other

Does your lab score CNVs differently if they are prenatal vs postnatal?

210 responses

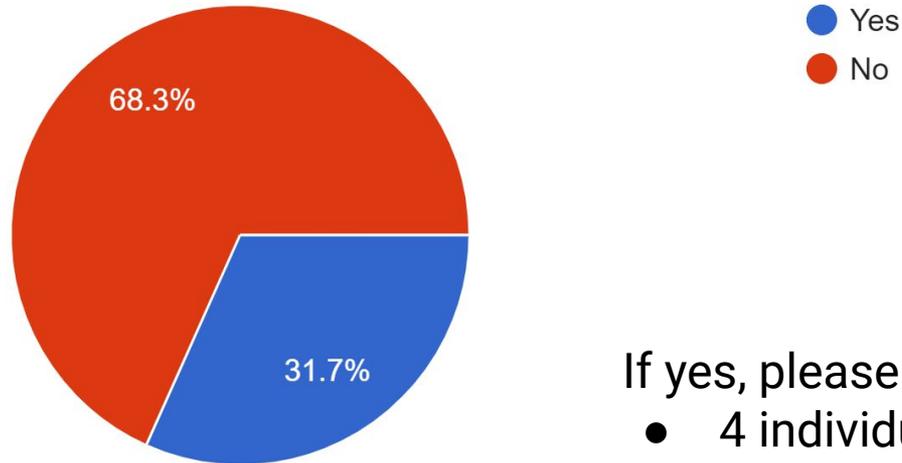


If yes, please describe:.

- 19 individuals provided comments on scoring differently in a prenatal setting: VUS reported less frequently, size cutoff greater

If you report AR gene deletions, do you report deletions focal to AR genes with high DGV coverage (such as NPHP1 or PARK2), if there are no clinical notes on the patient or if the notes are inconclusive?

205 responses

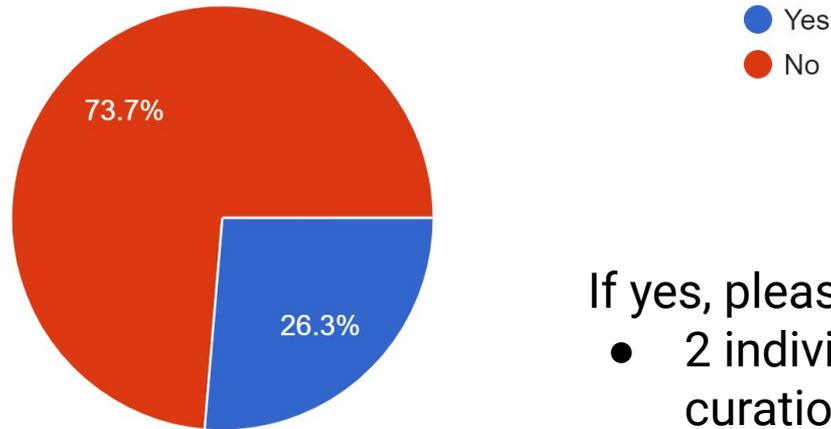


If yes, please describe:

- 4 individuals commented on reporting these as carrier status
- 2 individuals note that they discuss these on a case by case basis

Do you use scoring metrics for recurrent CNVs with reduced penetrance and variable expressivity (such as 15q11.2 NIPA 1/2 deletion)?

205 responses



If yes, please describe:

- 2 individuals utilize ClinGen Dosage curation
- 1 individual reports based on metrics with note
- 1 individual underscores these CNVs