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The Genetic Testing Reference Materials Coordination Program (GeT-RM) is a CDC initiative that aims to improve the availability of appropriate and characterized reference materials for genetic testing. We compared the gene panel test data submitted to the GeT-RM by clinical labs for reference sample NA12878, to the gold-standard data produced for this sample by the NIST hosted Genomes in a Bottle Consortium (GIAB). We also compared the results to those achieved with an augmented exome sequencing assay developed by our lab: the ACE Clinical Exome Test (Patwardhan et al. 2015).

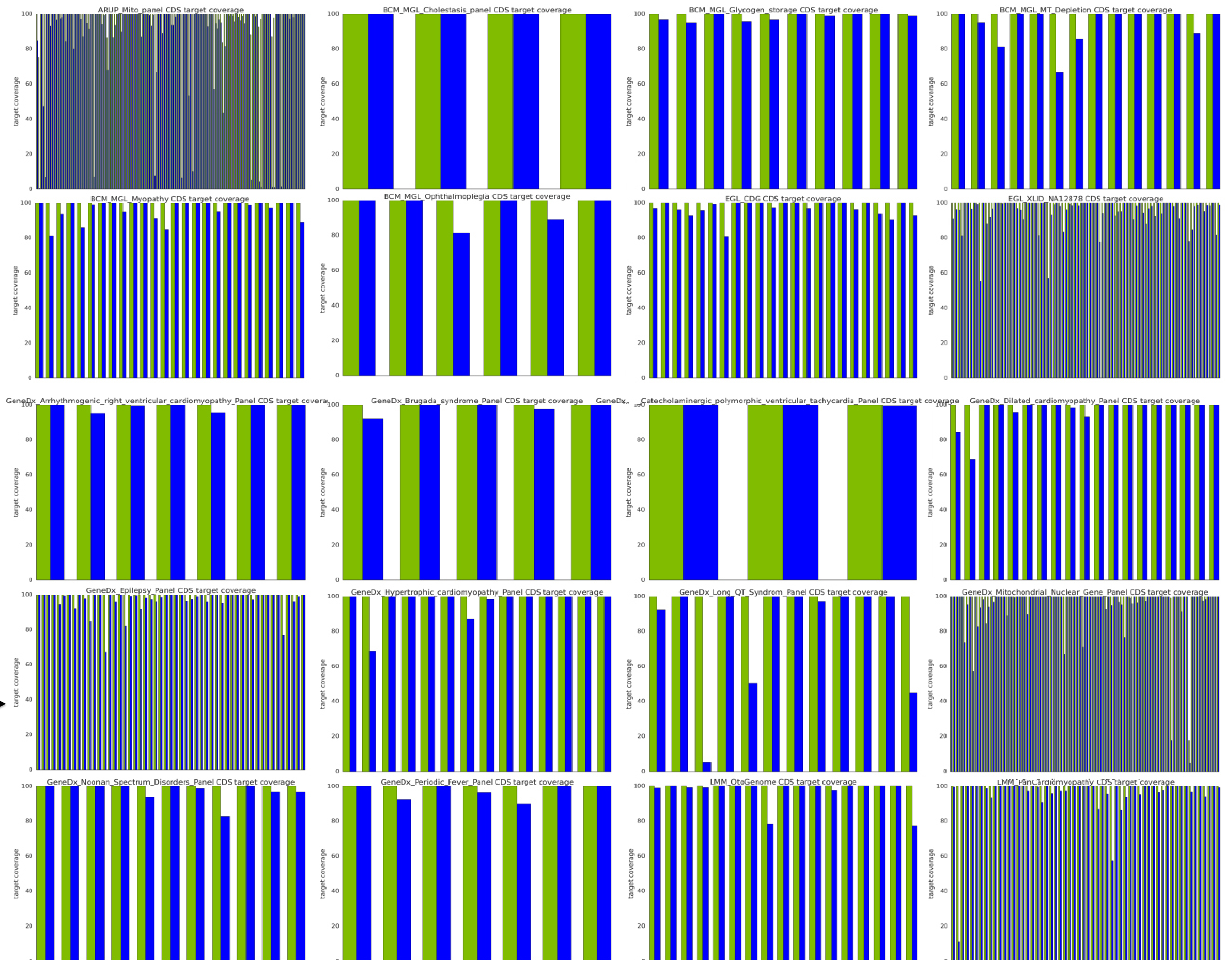
| Gene | ACE Clinical Exome Test (%) | Gene Panel Test (%) |
|-------|-----------------------------|---------------------|
| FXTN | 100 | 98 |
| P600 | 100 | 100 |
| ITGA1 | 100 | 90 |
| POMT1 | 100 | 100 |
| ITGA9 | 100 | 100 |
| ITGA2 | 100 | 98 |
| SERTF | 100 | 86 |
| SERN1 | 100 | 100 |
| POMT2 | 100 | 100 |
| POLA2 | 100 | 90 |
| POLA3 | 100 | 100 |
| POLA4 | 100 | 100 |
| APOE | 100 | 96 |

Panel genes ACEv3 performance

This histogram shows the distribution of finishing metrics for panel genes using the ACEv3 method. The x-axis represents the 'Finishing metric' from 0.3 to 1.0, and the y-axis represents the 'Number of genes finished' from 0 to 400. The data is highly concentrated at the maximum value of 1.0, with approximately 420 genes achieving this score. There are very few genes with scores between 0.3 and 0.8.

| Finishing metric | Number of genes finished |
|------------------|--------------------------|
| 0.3 | 0 |
| 0.4 | 0 |
| 0.5 | 0 |
| 0.6 | 0 |
| 0.7 | 0 |
| 0.8 | 0 |
| 0.9 | ~5 |
| 1.0 | ~420 |

The data for all gene panel tests pertaining to Mendelian disease diagnosis deposited in GeT-RM for reference sample NA12878, and for which GIAB data was available, were included in this analysis. Due to lack of GIAB reference data, two tests comprising entirely of the mitochondrial genome (MT) were excluded (GeneDX_NA12878_Mitochondrial_Genome & BCM_MGL_NA12878_Whole_Mitochondrial_Genome) and MT data was excluded from two further tests (GeneDX_NA12878_Dilated_cardiomyopathy_Panel & LMM_NA12878_OtoGenome). Two panels relating to somatic variant detection and drug metabolism were also excluded (GPS_WUSTL_Cancer_panel & GPS_WUSTL_Drug_metabolism_panel).

[illegible]

1. Patwardhan et al. *Genome Medicine* 2015, 7:71
2. GeT-RM web site at NCBI: <http://www.ncbi.nlm.nih.gov/variation/tools/get-rm/>
3. GIAB reference: <http://www.ncbi.nlm.nih.gov/pubmed/24531798>