

HITSAC GENOMICS WORKING GROUP
CANDIDATE USE CASE: GENETIC TEST RESULTS REPORT FORMATS

On a CAP working group we found that genetic test results (i.e. the specific findings) are often reported in different formats driven by the technology used to identify the mutation.

For instance, many kits will not provide the specific mutation identified but 'roll-up' the result into biomarker which is associated with 1 to many mutations. Tests performed on ABI sequencing platform are often reported using HGVS nomenclature and no genomic coordinates. Tests performed on NGS platform are first defined in genomic coordinates and then translated into various formats including HGVS and corruptions thereof for reporting.

So how does one put together the genetic finding on a patient and how do you support analytics - decision support and panel management or population health?

NLM (Clem McDonald from the Lister Hill Center) NCBI and I tried to work with CAP to create a map of biomarkers and mutations or 'rules' for determining associated mutations, but this effort was sidetracked for other CAP priorities. We know how to do it and have HL7, NLM, and NCBI support but need to get an appropriate clinical partner.