Center for Genomic Interpretation

ELEVATEGENETICS LANDSCAPE

Technical billing accuracy analysis. For Research Use Only. Not for clinical use.

The test and lab details in this mock report are fictional yet are modeled after real clinical lab scenarios.

NPI #####

Test Name: Hereditary Cancer Panel

Laboratory: HEREDITARY TESTING EXPERTS, INC

Date: 17 January 2022

Test Description

Hereditary cancer screening. A genetic test using blood or saliva from a patient to sequence and report variants associated with increased incidence of cancer.

FINDINGS

BILLING FRAUD: Very Likely

The 81432 CPT code for the full gene sequencing of at least these 10 genes: BRCA1, BRCA2, CDH1, MLH1, MSH2, MSH6, PALB2, PTEN, STK11, and TP53, has been repeatedly used by the laboratory and reimbursed by [the Client]. However, data provided by the laboratory suggests that more than 10% of the time, the following genes are not fully sequenced in the exonic regions: BRCA1, PALB2, STK11 and TP53. **Further investigation is recommended.**

REPORTING PRACTICES: Possibly Misleading

The reports provided by the laboratory do not clearly list the regions that were not sequenced to sufficient depth for accurate reporting according to the laboratory's technical specifications. Manual inspection of intermediate files revealed that several regions did not have sufficient coverage for accurate reporting.

METHODOLOGY & RESULT DETAILS

Billing Fraud (CPT Code Usage)

The technical specifications for the test under investigation were obtained through CGI contacting the laboratory's Customer Service team. CGI also contacted the laboratory staff directly and asked for any regions of the assay that had dropped out more than 10 times in the last 100 clinical samples run on the assay. The genes and exons corresponding to these coordinates were analyzed by CGI for consistency with services that should have been performed based upon the billed CPT code.

Results on CPT Code Usage

CPT code 81432 has been used exclusively from this laboratory to bill for their hereditary cancer panel test. CMS' summary of this code is as follows: "HEREDITARY BREAST CANCER-RELATED DISORDERS (EG, HEREDITARY BREAST CANCER, HEREDITARY OVARIAN CANCER, HEREDITARY ENDOMETRIAL CANCER); GENOMIC SEQUENCE ANALYSIS PANEL, MUST INCLUDE SEQUENCING OF AT LEAST 10 GENES, ALWAYS INCLUDING BRCA1, BRCA2, CDH1, MLH1, MSH2, MSH6, PALB2, PTEN, STK11, AND TP53." All of these 10 genes are considered Loss of Function genes for the purpose of clinical interpretation, meaning that clinically significant regions include the exons of these genes (i.e. the part of the genes that code for its protein gene product). Clinically significant exon regions of the following genes dropped out of the assay for more than 10% of patients on these genes for this laboratory: BRCA1, PALB2, STK11 and TP53. This is not consistent with the CPT code being used.

Reporting Practices

In order to ensure that reporting practices appropriately represent any gaps in sequence coverage, CGI asked the laboratory to sequence and provide intermediate sequencing analysis data files (BAMs) for three NIST reference standards (non-patient samples). In addition to providing files related to the analysis, CGI also asked the laboratory to issue a report for the samples. Coverage data from BAM files for these three references was manually inspected for consistency with regions of low coverage listed on the final report (if any).