September 27, 2022

Submitted via healthit.gov/isa/uscdi-data-class/genomics

Re: USCDI v4 Comment Period

To Whom It May Concern,

Thank you for the opportunity to comment on data elements and classes that should be included within USCDI v4. Invitae Corporation would like to comment in support of increasing the Genomics Data Class to Level 2 and to request its inclusion within USCDI v4. Invitae is a leading medical genetics company, delivering genetic information services that support a lifetime of patient care — from inherited disease diagnoses and family planning to proactive health screening and personalized diagnosis, treatment and monitoring of cancer — combining genetic and clinical information to improve health decision-making. Our pursuit is to improve healthcare for everyone, including by making genetic testing more accessible and more affordable to all who may benefit. Throughout the company's lifetime, we have served over 3 million patients, providing them information relevant to their care for decades to come. Because genetic and genomic data have pertinent long-term uses, Invitae is a strong supporter of ensuring that genetic test results are incorporated into USCDI data standards, so that these data can follow patients across the many healthcare providers and systems they may interact with throughout their lives.

As we progress toward precision medicine, the use of genetic information in patient care is increasingly important. Genetic testing prices are decreasing and more patients are getting access to genetic information that can have utility throughout their lives. However, barriers remain that prevent patients and their clinicians from realizing the most value from this information — namely, lack of integration into electronic health records (EHRs). Invitae serves a large patient population and currently has to contract with individual EHR vendors or health systems to integrate our orders and results into a patient's EHR. This process is fragmented, time-consuming, costly, and does not ensure that a patient will retain easy access to their genetic testing results if they happen to change EHR vendors or health systems.

Without undergoing this costly process of EHR integration, Invitae's — and other laboratories' — genetic test results are attached to EHRs within a PDF. This makes it difficult for providers to find a patient's genetic test result and makes it virtually impossible to leverage clinical decision support tools. One case study where this lack of integration and easy access to genetic test results in lower utility of genetic information is in colorectal cancer. NCCN Guidelines were recently updated to recommend germline testing for all patients with colorectal cancer under the

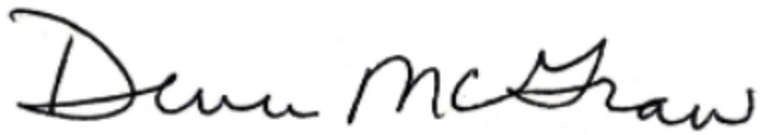
age of 50.[1] Germline testing can enable appropriate placement into clinical trials, identify eligibility for targeted treatments, and inform use of certain medications or surgeries to prevent secondary cancers, to name a few benefits. These benefits are more likely to be realized when the germline test results are in an accessible place in the EHR and physicians do not have to search for a PDF or rely on patient recollection to know if this information exists.

Inadequate standards for structured genomic data and lack of input from geneticists[2] were identified as major limitations preventing the easy access and use of genomic data within EHRs. The Electronic Medical Records and Genomics (eMERGE) Network showed that integration of genomic data within multiple EHR systems is possible and can enable more streamlined clinical decision support opportunities.[3] While the success of the eMERGE Project offers hope for better usage of genomic data within EHRs, Invitae believes that ONC, through USCDI v4, can accelerate the adoption of data standards conducive to increased utility of genetic test results.

For all these reasons, Invitae supports the elevation and inclusion within USCDI v4 of Level 1 Data Class: Genomics. Elevating this Data Class to Level 2 and incorporating it into the draft USCDI v4 will provide a necessary opportunity to receive stakeholder feedback on how to responsibly and appropriately integrate genetic data into electronic health records.

We appreciate your consideration of our comments. If you have any questions, please email me at deven.mcgraw@invitae.com.

Sincerely,



Deven McGraw
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[1] https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1436
[2] https://www.frontiersin.org/articles/10.3389/fgene.2019.01059/full
[3] https://pubmed.ncbi.nlm.nih.gov/31447099/