ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

January 4, 2021

Don Rucker, MD
National Coordinator for Health Information Technology
Office of the National Coordinator for Health Information Technology
Department of Health and Human Services
330 C Street SW
Washington, DC 20201

Re: Information Blocking and the ONC Health IT Certification Program: Extension of Compliance Dates and Timeframes in Response to the COVID–19 Public Health Emergency (RIN 0955–AA02)

Dear Dr. Rucker:

On behalf of the American College of Medical Genetics and Genomics (ACMG), we appreciate the opportunity to provide comments for consideration in response to the Information Blocking and the ONC Health IT Certification Program: Extension of Compliance Dates and Timeframes in Response to the COVID-19 Public Health Emergency interim final rule (RIN 0955–AA02). ACMG is the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics, and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. ACMG is the largest membership organization specifically for medical geneticists, providing education, resources, and a voice for more than 2,400 clinical and laboratory geneticists, genetic counselors, and other healthcare professionals, nearly 80% of whom are board-certified in the medical genetics specialties. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health.

ACMG supports ONC's decision to extend the information blocking applicability dates in response to the COVID-19 pandemic. However, COVID-19 cases are the highest we've seen since declaration of the public health emergency, and physician practices throughout the country are being significantly impacted. Even with the delayed compliance date of April 5, 2021, implementing changes to comply with the information blocking rule during the height of a public health emergency may

7101 Wisconsin Avenue
Suite 1101, Bethesda, MD 20814
Telephone: 301-718-9603
Fax: 301-718-9604

www.acmg.net

not be feasible for many practices.

Additionally, and of serious concern to the ACMG, there is much uncertainty about how to appropriately apply the preventing harm exceptions in §171.201, especially for the disclosure of genetic and genomic test results that, in general, should be delayed for the purpose of preventing patient harm. **For these reasons, we urge ONC to delay the applicability date of the final information blocking rule until agency-level guidance is released to assist healthcare providers and clinical testing laboratories on complying with the final rule while also balancing the need to prevent patient harm.**

Application of the Preventing Harm Exception

ACMG is concerned about the potential harm to patients that may occur if genetic and genomic testing results are automatically released to patients. We appreciate inclusion of the preventing harm exception in §171.201 and anticipate that this exception will be frequently used for return of certain genetic and genomic testing results. However, we remain concerned about the focus on patient-specific exceptions rather than institutional-level exceptions, at least for certain types of genetic and genomic results. The preamble to the final rule (85 FR 25642, pg. 25840) explains that exceptions to prevent harm could be met in two ways: *through a ''qualifying organizational policy'' (§ 171.201(b) as proposed)* ***OR*** *through a ''qualifying individualized finding'' (§ 171.201(c) as proposed).* However, the final language in §171.201 makes it clear that organizations must comply with both §171.201(b) **AND** at least one of the two provisions in §171.201(c) which state that the risk of harm must:

> *(1) Be determined on an individualized basis in the exercise of professional judgment by a licensed health care professional who has a current or prior clinician-patient relationship with the patient whose electronic health information is affected by the determination; or*
>
> *(2) Arise from data that is known or reasonably suspected to be misidentified or mismatched, corrupt due to technical failure, or erroneous for another reason.*

There are numerous ways in which automatic release of genetic and genomic results could lead to patient harm, such as with certain unanticipated findings. Such scenarios do not appear to meet the criterion in §171.201(c)(2) nor are they events that can be anticipated on an individual basis as described in §171.201(c)(1). The potential for harm from automatic release of certain genetic and genomic information may be universal and should not require individual determinations, and **clarification from ONC is needed to ensure that delay of results policies intended to prevent patient harm are implemented appropriately.**

Clarifications around appropriate use of the preventing harm exception are particularly important for external clinical testing laboratories. Under the new rule, the laboratory is an "actor" responsible for releasing results to the patient. In the preamble to the final rule, in response to a public comment specifically about release of genetic results, ONC explains that the ordering clinician would be well

positioned to make individual determinations consistent with §171.201(c)(1) as to when the patient would be at risk of harm from automatic release of test results (85 FR 25642, pg. 25843). However, this assumption is not necessarily true for the genetic testing laboratory which is responsible for releasing results to the patient but generally is *not* positioned to make such individualized determinations. In addition, very often, the ordering physician is not sufficiently trained in genetics to make individualized determinations regarding the risks of release of test results, and consultation with a medical genetics professional may be needed. Since individualized determinations cannot always be made by the laboratory, institution-level laboratory policies to automatically delay release of certain types of results are necessary to allow the ordering physician an opportunity to provide a diagnostic interpretation and arrange for counseling if needed.

Examples of Potential Patient Harm

We agree, of course, that patients should have access to their genetic and genomic data; however, the timing of release of such information must be considered in light of the need for interpretation by the treating physician, often in consultation with a medical geneticist or a genetic counselor, to prevent patient harm. Standard practices include delaying release of certain potentially alarming genetic results, such as those for Huntington disease, by at least a few days to provide the physician time to review the results and communicate them to the patient and/or arrange for the patient to have a return of results session with a medical geneticist or genetic counselor. Additionally, routine genetic testing in clinical practice often includes results of uncertain significance or carrier status that could easily be misinterpreted by the patient.

More complex genomic results may require a longer delay, providing the physician an opportunity to clarify and interpret the results and consider them in light of other patient medical information. These laboratory results may not be misidentified or erroneous as covered in §171.201(c)(2); they just cannot be considered complete until the physician interpretation is included. As noted in *The interface of genomic information with the electronic health record: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG)*[1], the clinician's interpretation provides information that clarifies and could alter significantly the interpretation of the raw genomic data provided in the laboratory report. Therefore, the genomic dataset should not be made available to the patient until the clinical information is included to complete the diagnostic interpretation. For particularly complex results, coordination among multiple providers (e.g., clinical geneticist, laboratory geneticist, and other medical specialist such as a neurologist) may be necessary to address the complexity of analysis, interpretation, and patient communication.

Further, while patients can be pre-counseled for potential anticipated findings, genetic and genomic testing may uncover unexpected secondary findings. In some cases, it may be the patient's preference to not receive certain types of secondary findings such as those related to late-onset conditions for which there is not a therapeutic intervention that would alter or prevent onset. As another example, a patient/couple undergoing prenatal testing may not want to know the gender of

[1] Grebe TA, et al; The interface of genomic information with the electronic health record: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 2020; 22:1431–1436.

the fetus. In this case, the laboratory is required to provide the finding, but the ordering healthcare provider can ensure that this information will not be visible to the patient. Genetic and genomic tests can also reveal unanticipated findings such as misattributed parentage[2] or consanguinity[3] which are best delivered along with appropriate counseling services. In some cases, genetic testing could reveal evidence of abuse for which the healthcare provider has a duty to report in accordance with state or local laws[4]. Unanticipated findings such as these may only be evident to the treating healthcare provider who has additional information about the patient, which is another example of why general delay policies are needed for the laboratories responsible for releasing genetic test results to patients.

Conclusions

Institution-level delay of return of certain genetic and genomic test results is an important process that minimizes patient harm from direct access to genomic data prior to provider review, interpretation, and communication with the patient. However, there is much uncertainty regarding whether policies that include such embedded delays are in compliance with the new rule. **We urge ONC to delay the applicability date of the final information blocking rule until agency-level guidance is released to assist healthcare providers and clinical testing laboratories on complying with the final rule while also balancing the need to prevent patient harm.** The ACMG is prepared to work with ONC to identify the types of questions and clarifications that could be addressed through additional agency-level guidance.

For additional information, please contact Dr. Michelle McClure, ACMG Public Policy Director, at mmcclure@acmg.net.

Sincerely,

Anthony R. Gregg, MD, MBA, FACOG, FACMG
President
American College of Medical Genetics and Genomics

Maximilian Muenke, MD, FACMG
Chief Executive Officer
American College of Medical Genetics and Genomics

[2] Deignan JL, et al. Points to consider when assessing relationships (or suspecting misattributed relationships) during family-based clinical genomic testing: a statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 2020; 22:1285–1287.
[3] Rehder CW, et al; American College of Medical Genetics and Genomics: standards and guidelines for documenting suspected consanguinity as an incidental finding of genomic testing. *Genet Med* 2013; 15:150–152.
[4] Sund KL and Rehder CW; Detection and reporting of homozygosity associated with consanguinity in the clinical laboratory. *Hum Hered* 2014; 77:217–224.