ACMG
American College of Medical Genetics and Genomics
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July 18, 2024

The PLUGS Medical Necessity Working Group

Re: ACMG Comments on the PLUGS Draft Consensus Recommendations and Framework for Development of Payer Medical Necessity Policies

Dear Working Group Members,

On behalf of the Economics of Genetic Services Committee of the American College of Medical Genetics and Genomics, we are providing the following feedback on the PLUGS Consensus Recommendations and Framework for Development of Payer Medical Necessity Policies (https://www.schplugs.org/wp-content/uploads/DRAFT-Consensus-Recommendations_Medical-Necessity-policies-05.20.24.pdf) which addresses the important problem of medical necessity in clinical genetic testing. First, we congratulate your group on developing the concept of this document and the thoughtfulness with which it is prepared. We unequivocally endorse the need for improved communication between stakeholders and the need for standards in demonstrating medical necessity. Diagnostic laboratories, patients, physicians, and other healthcare professionals all need greater predictability in decision making from health systems and payers in this emerging area of practice. Figure 2 is a good depiction of the general challenge with finding the right balance between restriction and flexibility, and could apply equally well in describing the evolution towards self-driving cars and the varying regulatory permissiveness of such technology on the roads.

We have a few points of feedback for your consideration:

A. There were many comments from the committee about specific aspects of evidence for medical necessity.
    1. The recommendations could be even more explicit about how evidence for medical necessity is organized and presented. The standards

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embodied in STARD[1] could be a starting point. Study designs that take into account the particular challenges of rare disease are needed.

2. A key issue revolves around what constitutes clinical utility of a particular service. The parameters of how clinical utility is demonstrated should be the focus of discussion and of study designs. ACMG has previously published a statement regarding expanding clinical utility. We suggest that we can collaborate to develop standards for various genetic conditions.
3. Regarding rare diseases and evidence for utility, are N-of-1 studies[2] being considered or could they be?
4. We believe that professional societies should be directly involved in the establishment of guidelines as standards for clinical utility to be used by payers. In the case of genetic diseases, the low prevalence of many disorders along with inadequate data on penetrance and expressivity, the continuing identification of new disease genes and variant interpretation, and evolving genomic technology all pose challenges in guideline establishment.
5. We do not anticipate a one size fits all strategy for the development of testing guidelines given the range of genetic disorders. For example, common diseases can be studied using a case-control approach which is not possible for rare diseases. For this reason, we propose that collaboration between payers, clinical geneticists, and laboratories should be encouraged to come to consensus for different categories of disorders. Some disease categories may require different approaches.
6. A well-written, evidence-based policy should have frequency limitations that are clinically logical and not burdensome on patients or healthcare

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[1] Cohen JF, Korevaar DA, Altman DG, Bruns DE, Gatsonis CA, Hooft L, Irwig L, Levine D, Reitsma JB, de Vet HC, et al. STARD 2015 guidelines for reporting diagnostic accuracy studies: explanation and elaboration. BMJ Open. 2016;6:e012799. doi: 10.1136/bmjopen-2016-012799

[2] Fountzilas E, Tsimberidou AM, Vo HH, Kurzrock R. Clinical trial design in the era of precision medicine. Genome Med. 2022;14:101. doi: 10.1186/s13073-022-01102-1

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professionals. In some cases, it is more cost-effective to repeat a test than to find and rely upon an older similar test. For example, a newer molecular test may be less expensive and more robust than a prior similarly named test. And, despite efforts to improve interoperability, the current reality is that finding accurate older test reports is sometimes very difficult and resource intensive.

7. We advocate for development and validation of research tools to perform the cost-benefit analysis on the problem of repeated genetic testing.
8. We agree that there are substantive differences between "standard of care" and "evidence-based," and we encourage further studies to fill the knowledge gaps for rare diseases.

B. We advocate for a shared responsibility to support evidence development. Payers have certain resources and expertise, particularly claims data, that can be useful in addressing key issues like appropriate utilization, underutilization, and longitudinal outcomes.

C. Where possible the recommendations could be described in simpler language, or where there are phrases that have a particular meaning in the field, these could be defined for lay stakeholders.

D. The use of supervised AI should be addressed. AI might be useful in addressing the problem of finding the appropriate policy relevant to a specific test.

E. Regarding "medical necessity policies that allow a broad path of reasonable care," we are concerned that "unlikely to cause harm" is not a sufficient standard. We advocate for an approach that accommodates testing that has demonstrable clinical utility.

F. We agree with the concept of using "wider guardrails for patients with multiple diagnoses and comorbidities" and note that this could be suitable to other patients with common clinical diagnoses where care is separated by long intervals and/or provided by different physicians.

G. Regarding the need for regular updates of policies, we note that CAP requests clinical laboratories review policies and procedures at least every two years. The CAP checklist is an example of a process that allows versioning. We are concerned

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that annual updating of policies would lead to increased costs for both labs and payers.

H. We advocate for evaluation of novel procedures where clinicians could provide additional clinical context and justification for the services requested.

I. We note that there is variability in test offerings from laboratory to laboratory; e.g., laboratories offering extended gene panels. We advocate for standardized base test content from which laboratories may extend and differentiate themselves.

J. We suggest further clarification of the use of ICD-10 codes in medical policies. Would a list include only covered codes or also commonly denied codes? We advocate for both transparency and flexibility.

K. Policies should provide a clear rationale for the medical necessity decision--allowing for patients, laboratories, and healthcare professionals to understand the rationale, be able to challenge the rationale (when appropriate), and plan for studies that could fill the evidence gap.

For questions or additional discussion, please contact Michelle McClure, PhD at mmcclure@acmg.net.

Sincerely,

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