

SACGHS Discussion of the Implications of Affordable Whole-Genome Sequencing

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Proposed Issues for Letter to the Secretary

- Challenges in evaluating the clinical validity and utility of WGS data
- Challenges in communicating WGS data to patients
- Coverage and reimbursement paradigm that does not meet the needs of WGS testing
- Timely and appropriate reassessment of WGS data as research reveals new findings
- Disparities and barriers to the equitable access to whole-genome sequencing (WGS) technologies (the meaning of “affordable”)

Proposed Guidance for WGS Issues

- Challenges in evaluating the clinical validity and utility of WGS data

Concern: limited information about clinical validity and utility for many associations and limited tools and resources for clinicians; current regulatory policy is not a good fit for whole-genome sequencing technologies

Guidance: HHS should apply the SACGHS oversight recommendations on clinical validity and utility to WGS technologies.

Proposed Guidance for WGS Issues

- Challenges in communicating WGS data to patients

Concern: determining if/when/how to communicate incidental findings, variants of unknown significance, off-target results to patients; assuring a knowledgeable workforce.

Guidance: HHS should support professional societies in developing appropriate guidelines and implement SACGHS recommendations for genetics education and training.

Forum: professional societies such as ACMG, NSGC

Proposed Guidance for WGS Issues

- Coverage and reimbursement (C&R) paradigm that does not meet the needs of WGS testing

Concern: the current C&R paradigm may not be adequate to cover the informatics costs for WGS or the cognitive services required of clinicians.

Guidance: HHS should assess the remuneration needs of laboratory professionals and clinicians who provide/use WGS tests.

Forum: CMS Medicare Evidence Development & Coverage Advisory Committee

Proposed Guidance for WGS Issues

- Timely and appropriate reassessment of WGS data as research reveals new findings

Concern: as research advances, WGS data will need ongoing reinterpretation; it is unclear who will be responsible for updating the meaning and significance of the data.

Guidance: HHS should support the development of tools and resources (e.g., alert mechanisms in EHRs) that help assure the interpretation of patient data is current.

Forum: Office of the National Coordinator for Health Information Technology advisory committees, professional societies

Proposed Guidance for WGS Issues

- Disparities and barriers to the equitable access to WGS technologies

Concern: an “affordable” genome will not be affordable and accessible to all.

Guidance: to assure equitable access to WGS technologies, HHS should assess the feasibility of using WGS as part of a public health mandate such as newborn screening.

Forum: the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children