

NHGRI Definition of ‘Genomic Medicine’

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Genomic Medicine: *An emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use.*

- This is a purposefully narrow definition focused primarily on delineating a clinical discipline (e.g., cardiology or neurology) rather than a broad, more basic science-oriented research field (e.g., vascular biology or neuroscience).
- By ‘genomic,’ NHGRI means direct information about DNA or RNA, putting the study of more downstream products derived from the genome (i.e., proteomics, glycomics, metabolomics, etc.) as outside our immediate, focused view of genomics and, therefore, genomic medicine. This is due both to NHGRI’s small size and to its unique, traditional focus on nucleic acids. Other, broader definitions are possible— but for NHGRI’s purposes, the further one gets from the analysis of nucleic acids, the less ‘genomic’ the area tends to be.
- NHGRI recognizes that the dominant portion of its current mission and portfolio appropriately supports the foundational research that will ultimately produce the robust and vibrant discipline of genomic medicine. Such science is rapidly broadening beyond the study of genome structure and function to investigate the genomic basis of disease. The latter is immediately proximal to genomic medicine (as defined above), but increasingly reflects a central aspect of NHGRI’s research endeavors. An ideal term for this critically important area of human disease-oriented genomic research has not yet emerged, but is imperfectly captured by phrases such as ‘medical applications of genomics,’ ‘medically relevant genomic science,’ ‘genomic medical science,’ and ‘translational genomics.’
- In terms of the five research domains that provide the framework for NHGRI’s 2011 strategic plan for genomics, the fourth and fifth domains (‘Advancing the Science of Medicine’ and ‘Improving the Effectiveness of Healthcare’) capture the research activities falling under the umbrella of genomic medicine, as defined above. The other three domains are clearly essential to provide the foundational scientific basis for the research leading to genomic medicine rather than actually reflecting genomic medicine itself. Of particular note, the third domain (“Understanding the Biology of Disease”) largely captures the aforementioned human disease-oriented genomic research.
- Based on the above, genomic medicine can metaphorically be viewed as a key ‘destination’ for attaining NHGRI’s mission— improving human health through genomics research.