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PCV3 Global Landscape of Clinical Genomic Sequencing: Who, Where, and How?

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Abstract

This is a novel study of the global “landscape” of implementation of clinical sequencing (next-generation sequencing “NGS”). NGS is being adopted in several clinical areas, but adoption varies across countries and clinical conditions. We summarize current and projected clinical implementation across countries using structured reviews and interviews, and explore why these changes are taking place and future developments. We focus on the emerging use of exome and genome sequencing and three current uses of NGS: tumor sequencing, testing for rare and undiagnosed diseases (RUGD), and non-invasive prenatal testing (NIPT). We examine three topics: - Availability, utilization, and estimated expenditures for NGS across countries and regions - Labs offering NGS testing and illustrative test prices - Payer coverage of NGS tests and illustrative reimbursement rates. This study was conducted by the Global Economics and Evaluation of Clinical Genomics Sequencing Working Group (GEECS).