

Supplemental Online Content

Phillips KA, Douglas MP, Marshall DA. Expanding use of clinical genome sequencing and the need for more data on implementation. *JAMA*. Published online October 26, 2020.
doi:10.1001/jama.2020.19933

eTable 1. Next-generation Sequencing (NGS) Test Applications and Examples of Key Current and Emerging/Future Tests

This supplemental material has been provided by the authors to give readers additional information about their work.

eTable. Next-generation Sequencing (NGS) Test Applications and Examples of Key Current and Emerging/Future Tests

Current test	Emerging/future uses
Risk assessment and disease screening	
Germline cancer risk testing (eg, hereditary breast and ovarian cancer, lynch syndrome)	Circulating tumor DNA (“liquid biopsy”) for early detection of cancer Healthy patient screening for wide range of disease risks (including monogenic risk or tests based on polygenic risk scores)
Reproductive health decision-making	
NIPT for common trisomy syndromes in fetuses	NIPT for expanded genetic abnormalities in fetuses (eg, rare autosomal aneuploidies, microdeletions, large copy number variants)
Carrier screening for specific recessive genetic disorders (eg, cystic fibrosis)	Expanded carrier screening for a wide range of recessive genetic disorders
Diagnosis of an existing condition	
WES or WGS for diagnosis of specific clinical presentations of suspected genetic diseases (eg, intellectual disability disorders or rare diseases) (multigene panels are also currently used)	WGS as first-line test for diagnosis for suspected genetic diseases broadly
Diagnosis of infectious diseases (eg, SARS-CoV-2, influenza, urinary tract infections)	
Prognosis for a diagnosed disease	
Variation testing for prognosis (eg, FLT-3 in acute myeloid leukemia)	WGS for comprehensive assessment of genomic variations in acute leukemias
Prediction/monitoring of treatment response or adverse events	
Comprehensive tumor sequencing (including circulating tumor DNA liquid biopsy) for therapy selection and monitoring of cancer treatment (multigene panels are also currently used)	WGS for comprehensive assessment of genomic variations across cancer types
Pharmacogenomics panels to target current drug selection	Preemptive pharmacogenomics panels to guide future drug selection

Abbreviations: NIPT, noninvasive prenatal testing; SARS-CoV-2, severe acute respiratory syndrome coronavirus 2; WES, whole-exome sequencing; WGS, whole-genome sequencing.