Data-Driven Medicine

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SOPHiA
GENETICS®

POLICY

Guidelines for diagnostic next-generation sequencing

Gert Matthijs[*,1,8], Erika Souche[1,8], Mariëlle Alders[2], Anniek Corveleyn[1], Sebastian Eck[3], Ilse Feenstra[4], Valérie Race[1], Erik Sistermans[5], Marc Sturm[6], Marjan Weiss[5], Helger Yntema[4], Egbert Bakker[7], Hans Scheffer[4] and Peter Bauer[6]

ARTICLE

A standardized framework for the validation and verification of clinical molecular genetic tests

Christopher J Mattocks[*,1,7], Michael A Morris[2,7], Gert Matthijs[3,7], Elfriede Swinnen[3], Anniek Corveleyn[3], Els Dequeker[3], Clemens R Müller[4], Victoria Pratt[5] and Andrew Wallace[6], for the EuroGentest Validation Group[8]

Molecular Biomarkers for the Evaluation of Colorectal Cancer

Guideline From the American Society for Clinical Pathology, College of American Pathologists, Association for Molecular Pathology, and American Society of Clinical Oncology

ACMG PRACTICE GUIDELINES | Genetics inMedicine

ACMG clinical laboratory standards for next-generation sequencing

Heidi L. Rehm, PhD[1,2], Sherri J. Bale, PhD[3], Pinar Bayrak-Toydemir, MD, PhD[4], Jonathan S. Berg, MD[5], Kerry K. Brown, PhD[6], Joshua L. Deignan, PhD[7], Michael J. Friez, PhD[8], Birgit H. Funke, PhD[1,2], Madhuri R. Hegde, PhD[9] and Elaine Lyon, PhD[4]; for the Working Group of the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee

ACMG STATEMENT | Genetics inMedicine

Laboratory and clinical genomic data sharing is crucial to improving genetic health care: a position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors[1]

GDPR compliance – enforceable from 25th May 2018

The **General Data Protection Regulation (GDPR)** (Regulation (EU) 2016/679) is a regulation by which the European Parliament, the Council of the European Union and the European Commission intend to strengthen and unify data protection for all individuals within the European Union (EU). It also addresses the export of personal data outside the EU. The GDPR aims primarily to give control back to citizens and residents over their personal data and to simplify the regulatory environment for international business by unifying the regulation within the EU.[1] When the GDPR takes effect, it will replace the data protection directive (officially Directive 95/46/EC)[2] of 1995. The regulation was adopted on 27 April 2016. It becomes enforceable from 25 May 2018 after a two-year transition period and, unlike a directive, it does not require national governments to pass any enabling legislation, and is thus directly binding and applicable.[3]

The **EU General Data Protection Regulation (GDPR)** is the most important change in data privacy regulation in 20 years - we're here to make sure you're prepared.

TIME UNTIL GDPR ENFORCEMENT UTC
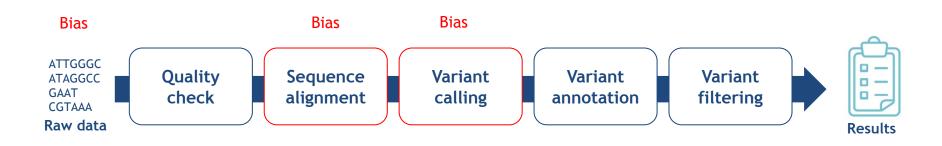**219:05:14:58**
Days   Hrs   Mins   Secs

Technical challenges in clinical routine

- Short turnaround time → Faster diagnosis

- No compromise between sensitivity and specificity → Accurate diagnosis

- Minimize regions to be analysed through alternative methods (Sanger, dHPLC, etc.) → Reduced costs

- Detect all types of variants:

    - Alterations in polyT/TG and GC-rich regions

    - Long tandem repeats

    - Copy number variations (CNVs)

    - SNVs, Indels

    - ALU repeats

Important to select the right technologies based on specific needs

Pre-validated tests – achieve top analytical performance

Bias   Bias   Bias

ATTGGGC
ATAGGCC
GAAT
CGTAAA

Raw data

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Quality check → Sequence alignment → Variant calling → Variant annotation → Variant filtering → Results
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Algorithm that adapts to each combination of

1. Sample type (blood, tumor,…)
2. NGS platform
3. Amplification/capture technology
4. Gene panel

Commercial Panels

Custom Panels

140+ employees

HQ: Lausanne, Switzerland

bsi.

ISO 13485
Medical Devices Quality Management

ISO/IEC 27001
Information Security Management