

GenomeMET: Metrology for genomic profiling to support early cancer detection and precision medicine

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Introduction

In Europe, cancer caused 1.26 million deaths in 2020 and the number is expected to increase in future. Genomic profiling diagnostic tools such as next generation sequencing are important in cancer treatment, particularly to develop personalised targeted therapy and to aid early diagnosis.

However, quality and comparability of genomic profiling from patient samples, including biobank samples, varies significantly with standards and metrological means to support the field being in their infancy.

Material & methods

GenomeMET, a project funded under the European Partnership for Metrology programme (22HLT06) comprising 17 partners from metrology institutes, sequencing centers and pathology & cancer institutes, focuses on the development of novel metrological capability, reference materials and reference measurement systems to support standardisation, regulatory compliance & quality/comparability of cancer genomic diagnostics.

Results/findings

GenomeMET uses tissue and liquid biopsy samples from lung and colorectal cancer as cancer models. Major outcomes of the project will be i) reference measurement systems to support validation, quality assurance and external quality assessment (EQA), ii) reference measurement procedures for cancer biomarkers and quality control parameters within genomic workflows, iii) assessment of existing and newly developed reference and EQA materials, and iv) a framework for measurement uncertainty determination.

Discussion/conclusion

Overall, this development of the needed metrological structure will help make genomic profiling for cancer prevention and treatment accessible across Europe and greatly contribute to the Horizon Europe's Mission on Cancer. Also, cancer research and biobanks using genomic profiling may benefit from more standardization in genomic profiling.

Key words: reference material, quality / quality control, sequencing, standardization, pre-analytics, cancer, regulatory compliance

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METROLOGY
PARTNERSHIP

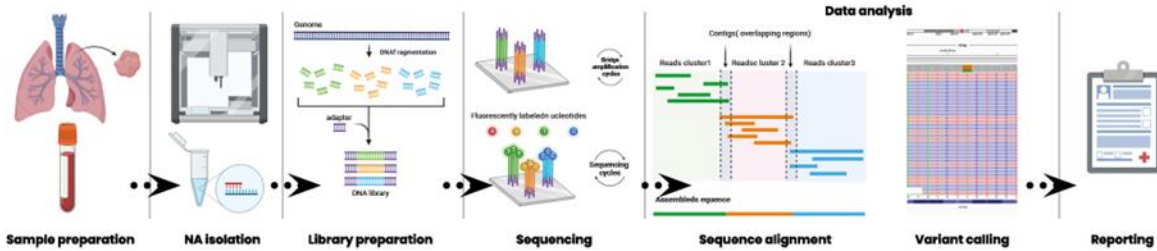


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GenomeMET: WP Structure

WP1: Reference Measurement Systems to support Genomic profiling

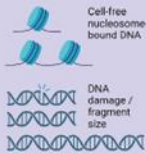


Lung & colorectal cancer models

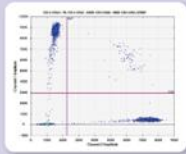
Tissue & Liquid Biopsy NGS workflows

WP2: RMPs:

High accuracy methods for NGS workflow quality metrics (e.g. NA quality/quantity)



SI-traceable dPCR measurement of key cancer biomarkers

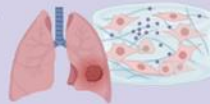


Preparation for JCTLM nomination & CCQM studies



WP3: RM/EQA materials

Novel RMs, EQA materials and "truth sets"



Framework for SI-traceable RM/EQA material value/identity assignment

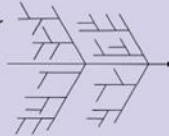


Framework for RM commutability assessment



WP4: MU & Multiparametric examinations

Statistical models for quantitative and qualitative genomic data uncertainty



Multiparametric data analysis

Framework for quantifying main contributors to uncertainty



WP5 Impact: Network of Excellence, novel metrology capability, RMS, Standards development, best practice guidance, training

WP6: Management and coordination

Project Partners

NATIONAL MEASUREMENT INSTITUTES



GENOMICS INSTITUTES



PATHOLOGY INSTITUTES



CANCER INSTITUTES



EQA PROVIDERS



TECHNOLOGY PROVIDERS

