

Integrated and STANdardized NGS workflows FOR Personalized Therapy

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Horizon 2020 - Work Programme 2018-2020 SC1-BHC-10-2019: Innovation Procurement: Next generation sequencing (NGS) for routine diagnosis

SCOPE

To implement **NGS in routine diagnostics** for personalised medicine and scale up demand-driven innovation for healthcare systems.

It should **lead to NGS tests, clinically validated procedures** (including sex analysis), **quality assurance schemes**, tools and methods for **data collection, management, analysis and interpretation**, with a view to **assist clinical decision making** and **foster medical research and innovation**.

Transferability and **cloud based NGS data analyses** should be considered, as appropriate.

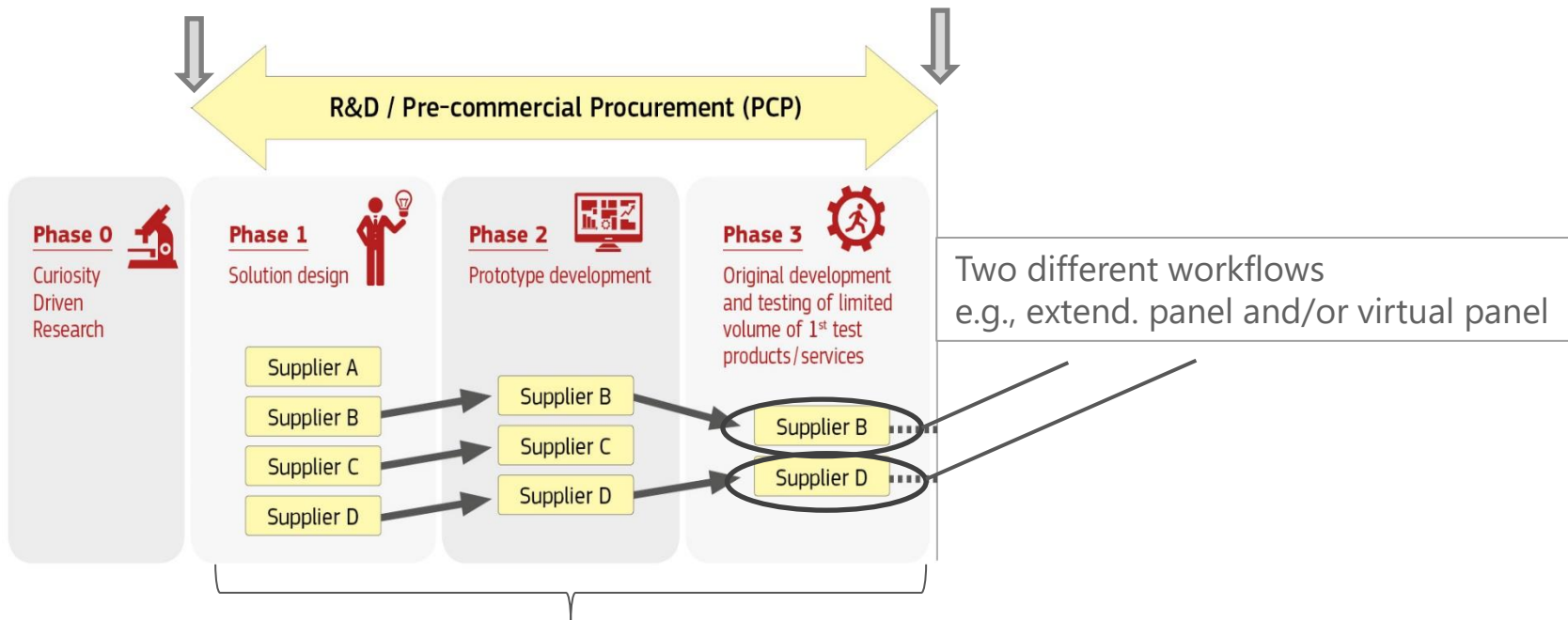
Input from initiatives like the EJP Cofund on **rare diseases and ERNs** should be considered when relevant. **Ethical issues** should be addressed.



The PCP Process

April 2022

April 2025
 (TRL 6-8)



ca. 2.5 Million €

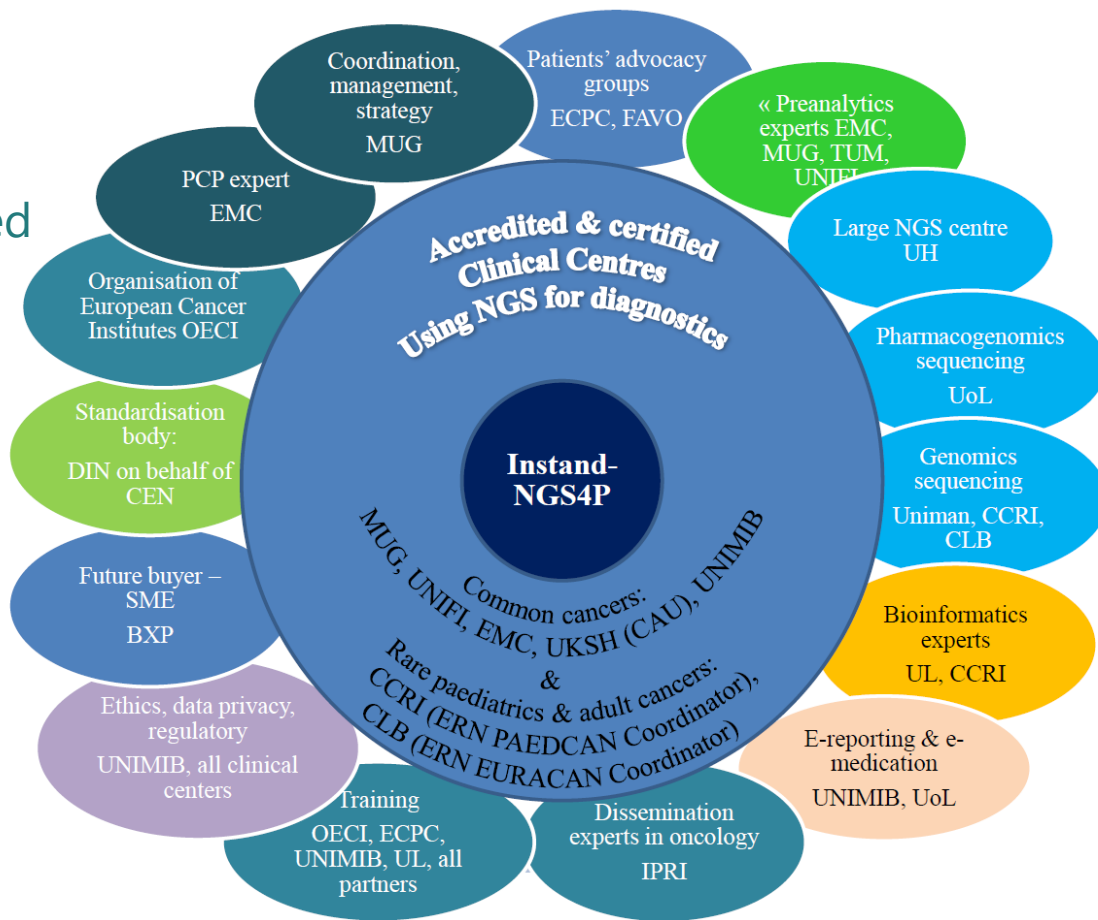
ca. 8.5 Million € including Buyer's contribution

Co-funded by the European Union

Grant Agreement n° 874719

18 Partners
 + > 200 associated
 medical centers

10 Countries



Specific Aspects to be Considered

- Application in routine diagnostics > requirements of IVDs (IVDR)
 - Role of standards
 - Need for reference material
 - Scientific validity, analytical and (clinical) performance
- Rare cancers may require „lab developed tests“
- WES or WGS as IVD > „virtual panels“
- Broad scope: workflow for different sample types; pharmacogenomics
- Integration of different solutions in a workflow

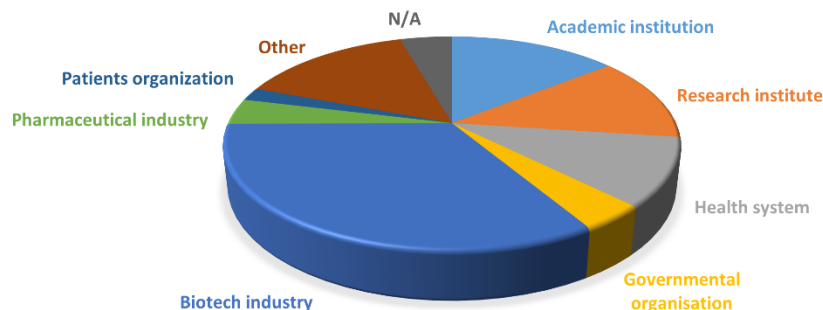


Lead: CCRI GmbH

Partners: all partners

Open Market Consultation

OMC activity
Virtual Meeting 22-23 March 2021
Webinars 28 April 2021 – Tendering process 18 May 2021 – Partnering
Questionnaires 17 May – 7 June 2021



Profile of external stakeholders registered in OMC events

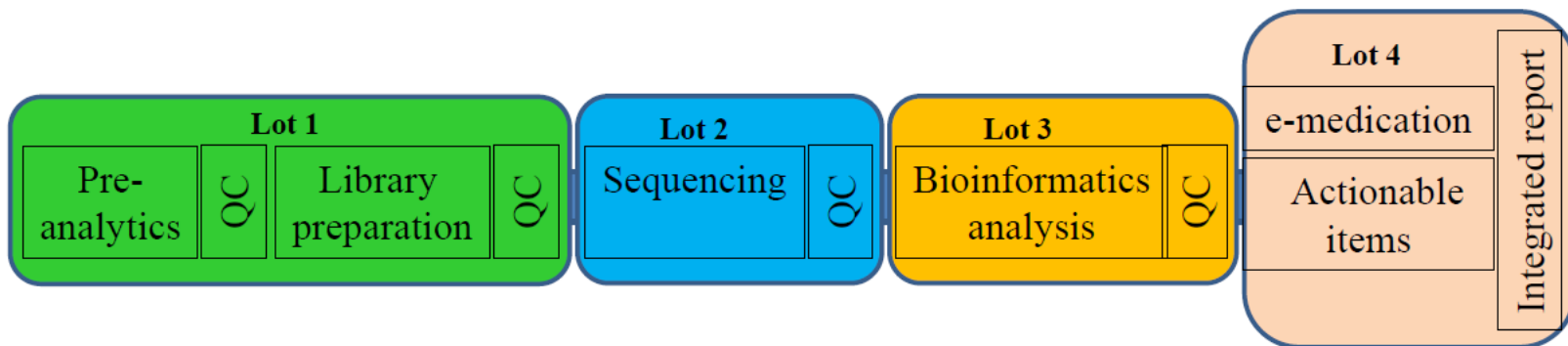
264 external participants (from 32 countries) took part in one or more OMC events

Participation from 89 companies



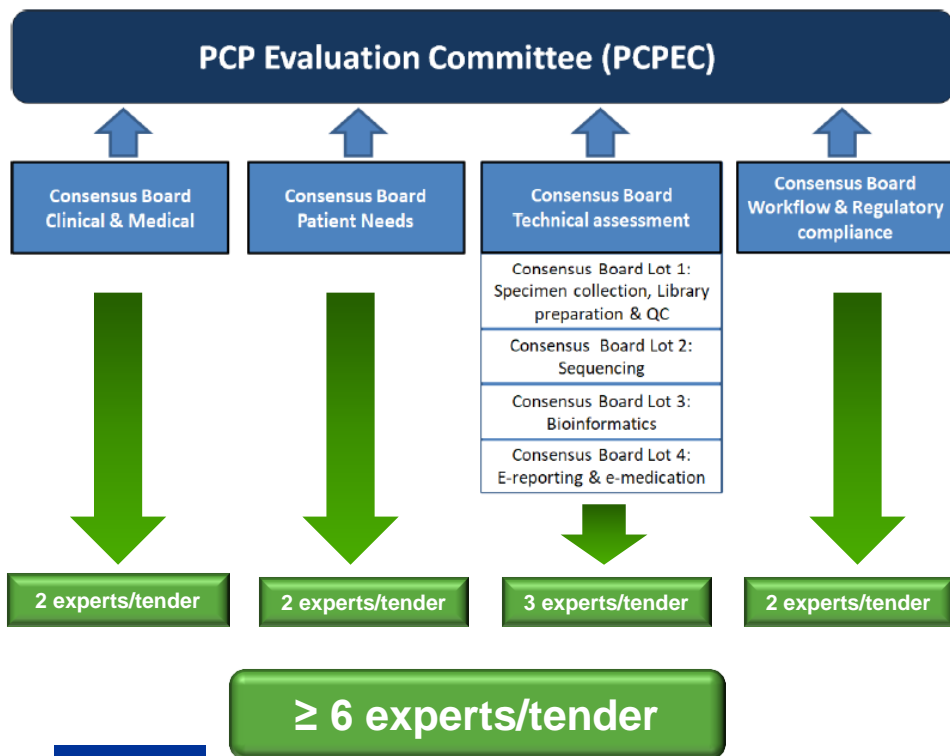
AN INTEGRATED AND STANDARDIZED NGS WORKFLOW

FROM PATIENT – TO - PATIENT



for common and rare cancers in adults and children

EVALUATION PROCEDURE



PCPEC: one representative from each group of Buyers

Consensus Boards: A list of experts was selected from among the project partners or external experts. The experts/members of the PCPEC had to report any potential Col.

- More than 30 experts were involved (1-5 proposals per expert).
- Each proposal was evaluated by at least 2 experts per consensus board (specific evaluation forms and dedicated online meetings, coordinated by Prof. Riegman).

Selection was based on the Evaluation Criteria:

- Exclusion Criteria
- Selection Criteria
- Award Criteria
 - Minimum Requirements
 - Weighted Award Criteria



Awarded Contracts for Phase 2

Lot	Phase 2 Ranking	Tender nr	Tenderer (Lead)	Tender Name
Lot 1:	1	1.5	Twist Bioscience Corporation	TwistPlatomicsMGI
	2	1.2	Integrated DNA Technologies, BV	IDT
	3	1.4	QIAGEN GmbH	Trinity
Lot 3:	1	3.9	Platomics GmbH	PlatoDKFZCharité
	2	3.4	BC Platforms AG	EU Onco-Platform
	3	3.3	Congenica Ltd	Congenica L3
	4	3.2	Fundacio Centre de Regulacio Genomica	Cancer Analysis
Lot 4:	1	4.2	Fundacio Centre de Regulacio Genomica	Cancer Reporting
	2	4.4	BC Platforms AG	EU Onco-Platform
	3	4.5	fragmentiX Storage Solutions GmbH	tde4ngs
	4	4.3	Congenica Ltd	Congenica L4

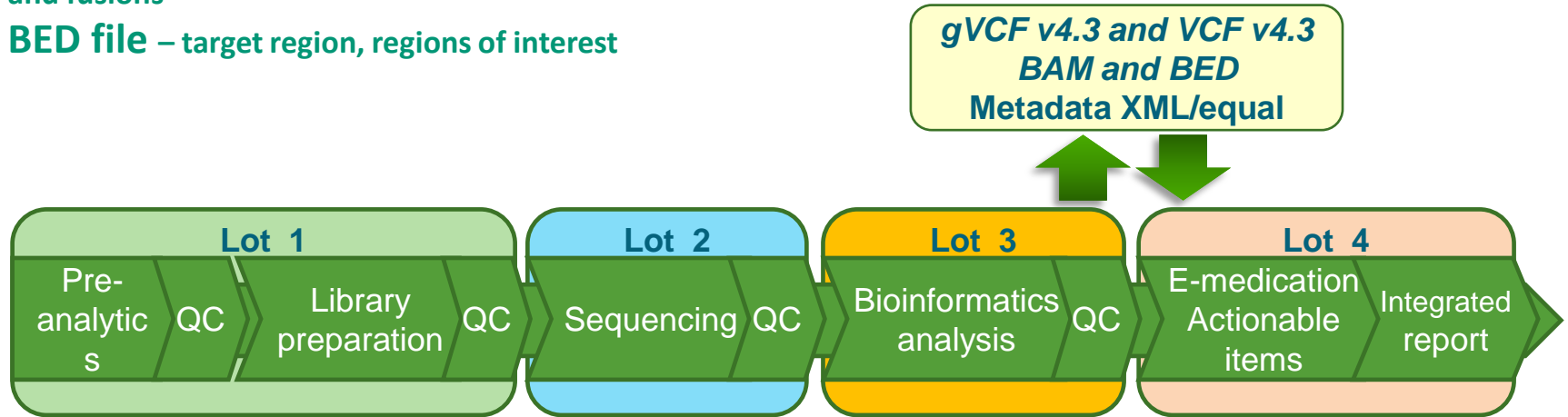
Some funding was used from Phase 3 to fund 4th contract for Lot 3 based on budget forecast for Phase 3



Lot 3 – Lot 4 Data Exchange

Variant specific formats:

- **gVCFv4.3 + VCFv4.3** - for coordinates for the variants: single nucleotides variants (SNV), Indels, Optional CNV and fusions
- **BED file** – target region, regions of interest



Contractors present:

Eufomatics, Platomics, Congenica, Fragmentix, CNAG

WP 7 Task 7.4

Lead: DIN Partners: all

- D7.6: Based on D7.5, a NWIP (DNA+RNA) was developed as input for standardization, **approved** by CEN/TC 140 and later split into **2 CEN/TS** to make the NGS workflow application more user friendly based on industry feedback:
 - WI 00140151, In vitro diagnostic Next Generation Sequencing (NGS) workflows — Part 1: Human DNA examination
 - WI 00140153, In vitro diagnostic Next Generation Sequencing (NGS) workflows — Part 2: Human RNA examination
- Both documents are very helpful for **guiding the R&D within Instand-NGS4P**
 - Total of 28 CEN/TC 140/WG 3 meetings



Useful Background Documents

D6.2 Dynamic informed consent matrix for NGS used in routine diagnostics UNIMIB Report Public

D6.3 Matrix for the minor's assent 6 - UNIMIB Report Public 24 D6.4 Guidelines to manage incidental findings for both patients and professionals 6 - UNIMIB Report Public

D6.5 Training program for genomic counseling 6 - UNIMIB Report Public



Conclusions

- Very broad scope of the project is a challenge
- PCP focus on SMEs. Very difficult for large companies to participate
- Exclusion of non-European sequencing platform providers
- Good collaborative spirit between contractors
- Major innovation expected:
 - Sample collection and stabilization (widely applicable workflow)
 - Improved bioinformatics pipeline (simple to use)
 - Integrated reporting for decision support at bedside
 - Information for patients
 - Advanced cybersecurity solutions
 - Solutions for CE-marked IVDs and LTDs



Many Thanks to The Team



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Thank you for your attention!

Please visit us at
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