

Paediatric Biomarkers and Biosamples Platform

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Why the need for paediatric biomarkers?

The majority of biomarkers developed in adults

Use not always safely extrapolated to children

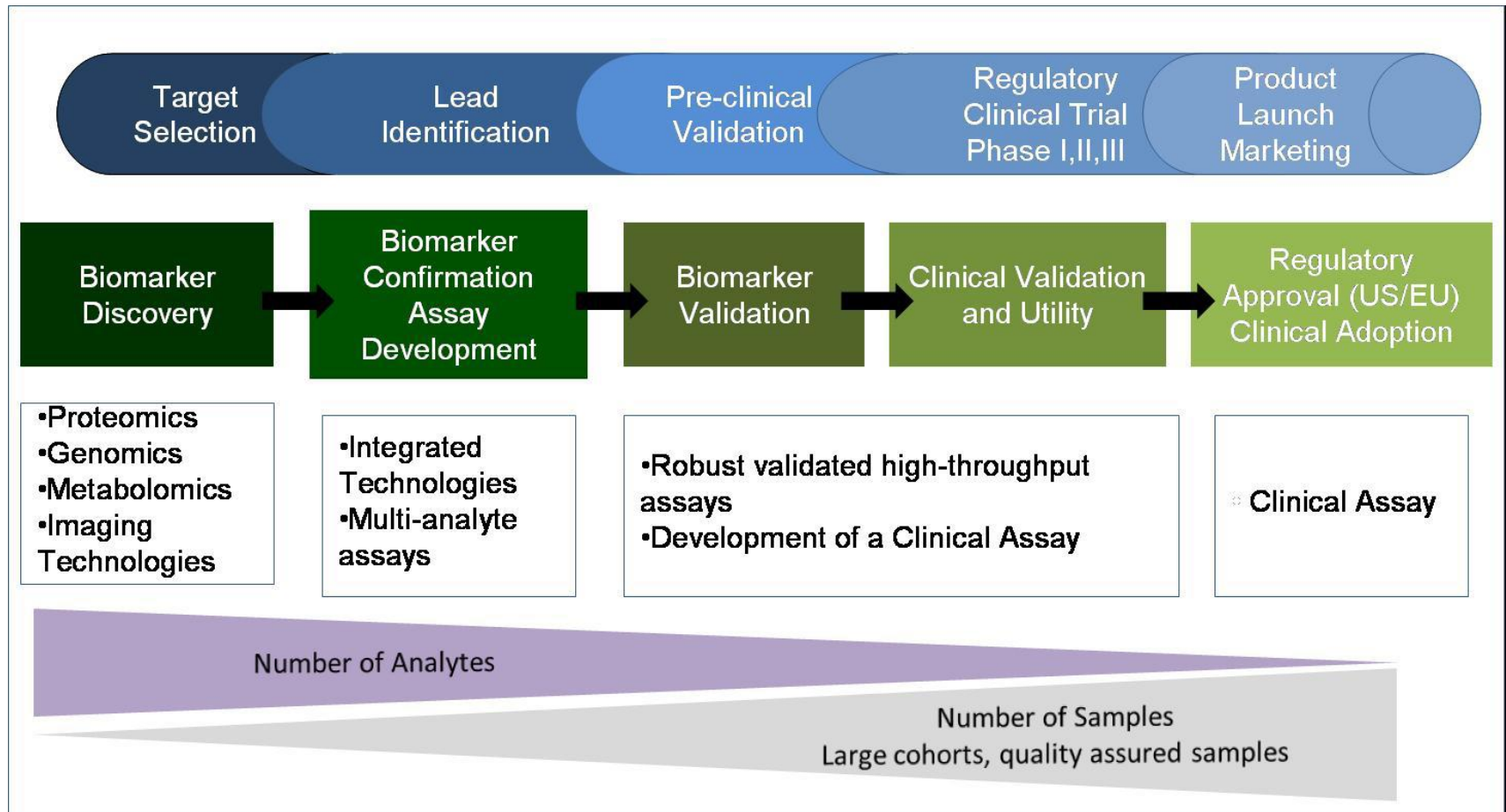
- Children pathogenesis may be different
- Some diseases unique to children
- Still growing and developing
- Gene expression can vary with age

Validated paediatric-specific biomarkers are vital

Paediatric biomarker research strewn with problems

- lower prevalence and incidence rates of disease
- heterogeneous populations
- more complicated bioethical considerations

Biomarker Discovery, Development and Validation



<http://education.molecularmedicineireland.ie/page/g/s/91>

Unavailable expertise/resources/facilities

Based on responses to EPTRI questionnaire (78 responders)

- Technical resources /laboratory facilities
- Access to high quality samples/biobanking
- Animal facilities
- Ethical and regulatory issues
- IT /biostatistics
- Omics platforms

Facilities are available...

Based on the same questionnaire

- identification/characterisation of biomarkers
- validation process of biomarkers
- biobank organisation including biosamples of paediatric interest
- Interest in making these available for a RI

Follow-up in-depth look at available capacity

22 responders in total (including 6 hospitals/clinics) in 11 countries

Omics platforms (7 countries)

- genomics (6), transcriptomics (6), metabolomics (3), proteomics (5)

Bioinformatics services (7 countries)

- 13 organisations

Biobanks and biosample provision (9 countries)

- 14 institutions (Mainly of serum, plasma and cells. Research and diagnostics.)
- Foetal, Preterm & Term new-born, Toddler, Children, Teenager, Parents (or family members)

Animal Models and Facilities (foetal and juvenile animals)

- Mouse, Rat, rabbit, fish and swine

Follow-up in-depth look at available capacity

In vitro models of human development

- embryonic stem cells (4), induced pluripotent stem cells (4), organoids (4)

Imaging of paediatric subjects/samples

- X-ray facilities (7), MRI facilities (9), CAT scan facilities (5), PET/MRI services (3). Databases of pathology images (7).

Techniques used for biomarkers identification and characterization

- protein arrays (7), tissue analysis (7), functional signalling pathway analysis (9), cytogenetic techniques (8), NGS (11), multiple secreted protein analysis (2), metabolic intermediate analysis (5)

Others

- Ethical and regulatory aspects related to biomarker validation for regulatory purposes (e.g. EMA qualification procedures)
- expertise on human data/biosample sharing for research activities
- compliance with GDPR

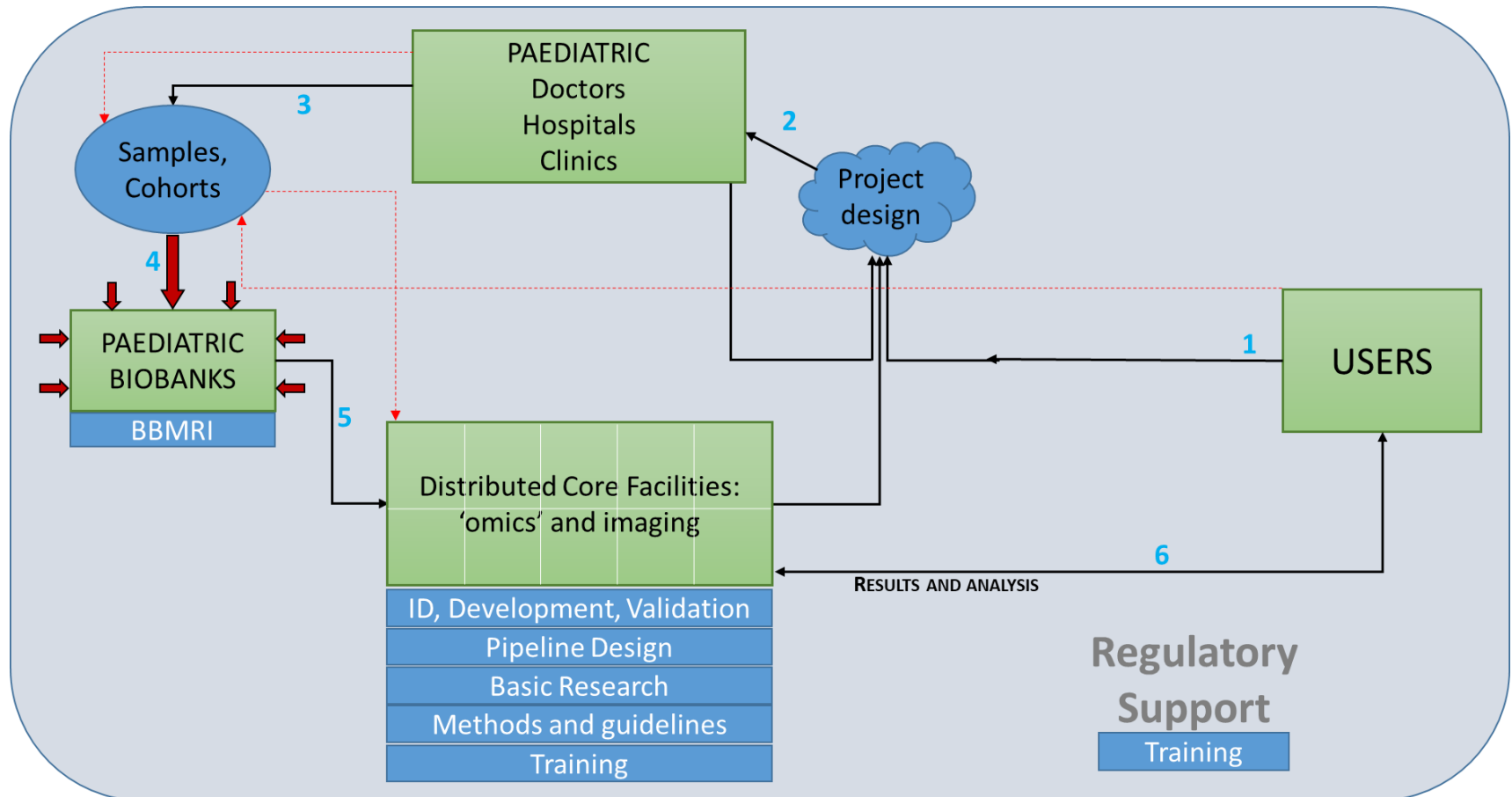
Services

To cover the conduct of studies aimed to the identification, characterization and validation of paediatric Biomarkers, the following services have been identified for provision within the platform:

1. Organisation and management of paediatric biosamples and related data for paediatric studies
 - ✓ Access to/deposit of annotated paediatric biological samples
2. Research activities aimed to identify biomarkers of high paediatric interest
 - ✓ RNA transcripts / DNA variants / Proteins / Metabolite candidate biomarker identification and characterisation in paediatric samples
 - ✓ Bioinformatics for the analysis of the data generated by omics platform
 - ✓ Verification in paediatric samples of the presence and levels of biomarkers (and candidate biomarkers; metabolites, proteins, RNA transcripts, DNA variants) previously identified in adult samples

**Trans-
national
access
activities
already
foreseen**

Biomarker and biosamples platform outline



Platform Partners

Partners

BBMRI-ERIC, CVBF , EATRIS-ERIC, CING, AP-HP, IPCZD, OPBG, UNN, SERMAS - HULP, TECHNION, UKR, ANTWERP, UZA

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