Omics to clinic

Goals:

Immediate

Integrate the multiple omics information to visualize expression levels in a more digestible form

Intermediate

Connect with clinical data. Apply analytics and predictive techniques.

Long term

Use in clinic for personalized medicine.

Basic Clinical thinking paradigm and workflow

What Happened to patient

Collect Info

- **History (pt / EMR)**
  - Onset/Course/ Frequency
  - Characterize/ severity
  - Events / Procedures
  - Family / Likelihood
  - Past EMR notes
  - OSH records

What's Happening to patient

Assess Info

- **Symptoms (pt / EMR / tests)**
- **Vitals / Exam /**
- **Labs / Imaging**

What We Think is happening to patient

Form / Narrow Differential diagnosis

- Patterns / Likelihood
- Test /Imaging results

What to do

Manage

- Rx / Tx / Surgery
- Algorithms
- Discuss / Consult
- Stabilize
- Prioritize

Investigate / Diagnose

- Testing / Imaging
- Trending

Anticipate / Prevent

- If / Then plans
- Thresholds
- Screen
- Prep
  - ex: type + cross

Check patient

Execute Plan

Clinical Genomics / Biomarkers

General Clinical Use Cases

1 **Disease Screening**
for Prevention / Early detection

2 **Predict Treatment Response**
Based on higher fidelity pathological classification

3 **Diagnosis**
Confirmation or rule out

4 **Rare Disease Diagnosis**
Exploratory

5 **Monitoring**
Recurrence and/or treatment efficacy

6 **Reproductive Risk Assessment**
Germline mutations

7 **Clinical Trials**
Eligibility & Enrollment

8 **Research**
Basic & Translational; Elucidating patients with disease-resistance

Clinical Genomics

Some example Specific Clinical Use Cases

Breast cancer HER2 + BRCA

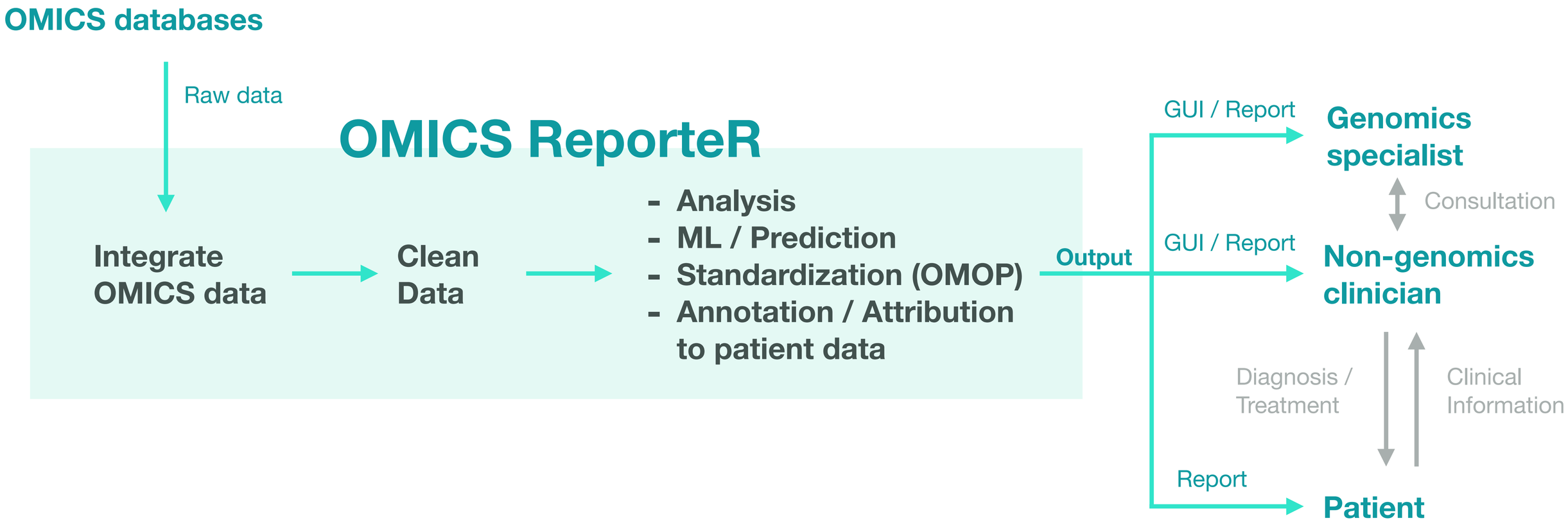
***MUTYH* colon cancer**

Prostate cancer

Warfarin
Sensitivity

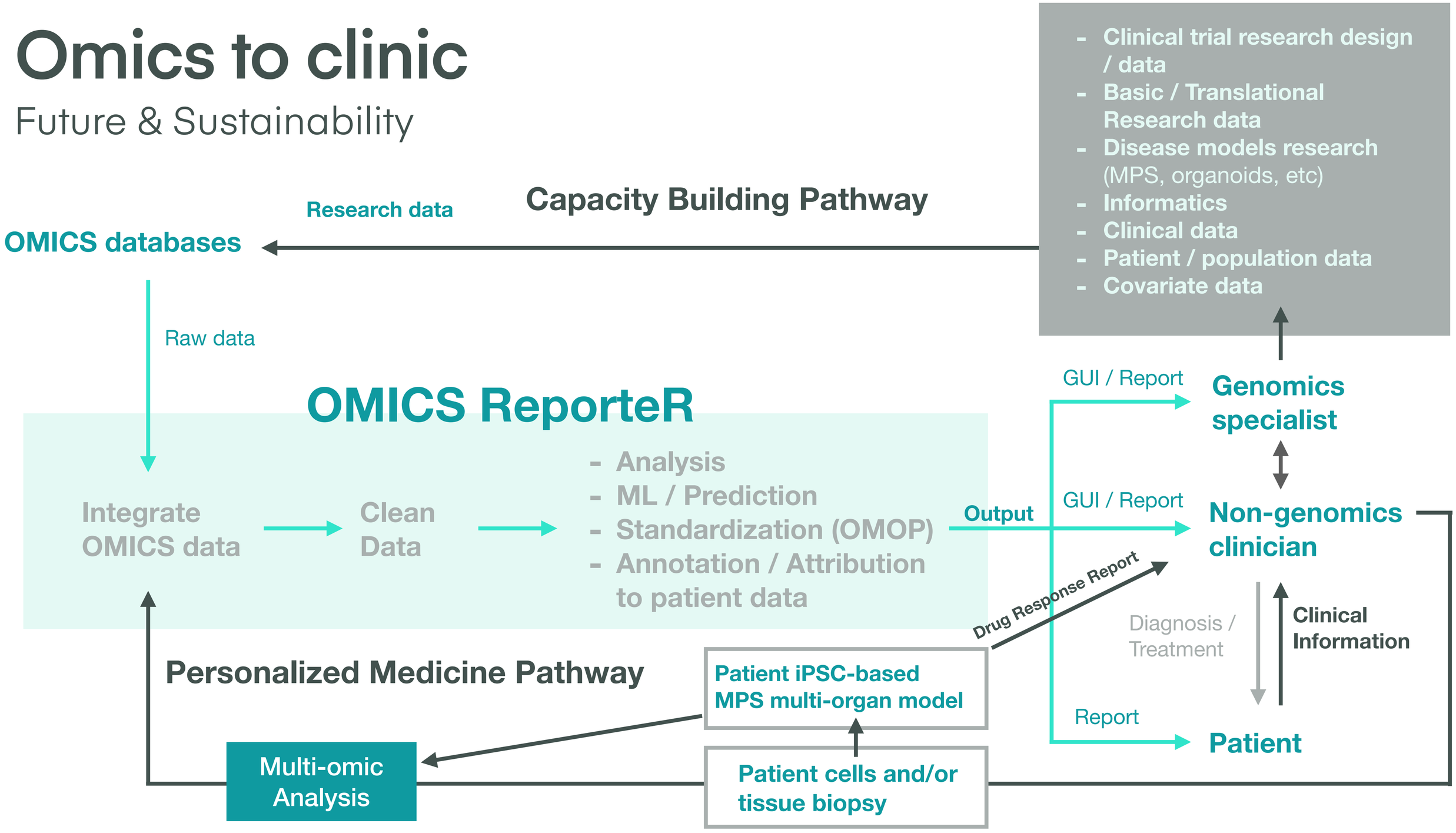
Omics to clinic

Pathway Schematic

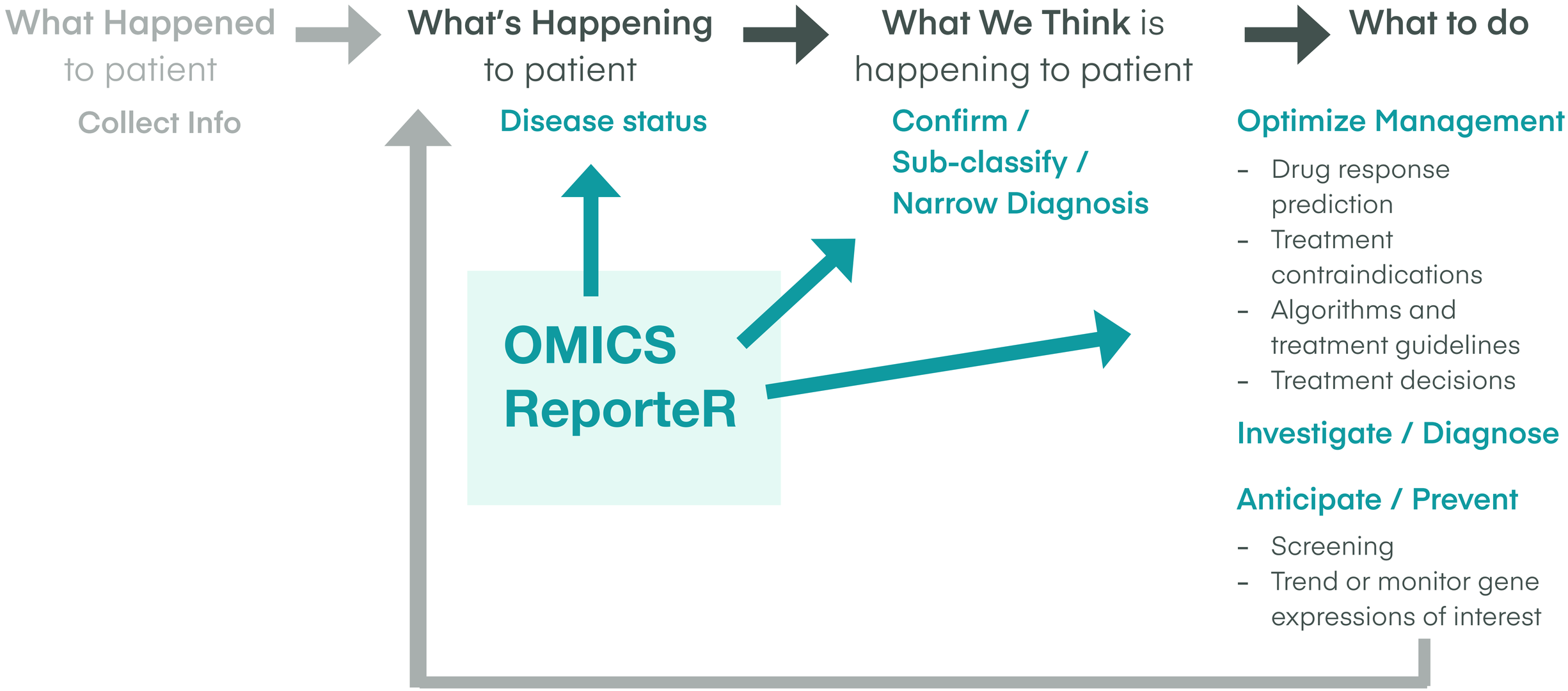


Omics to clinic

Future & Sustainability

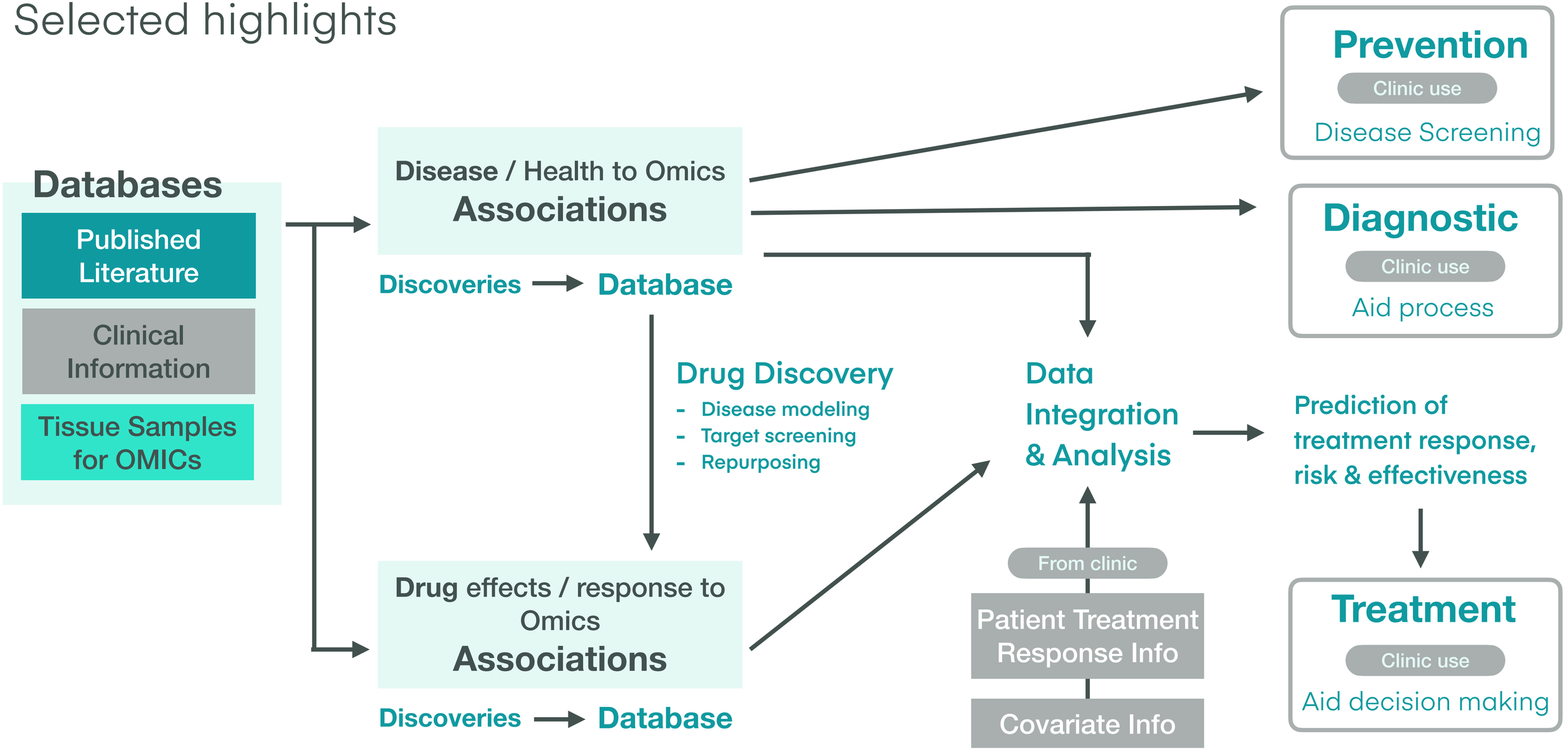


OMICS ReporteR potential areas of contribution



Technology progression for omics use in clinic

Selected highlights



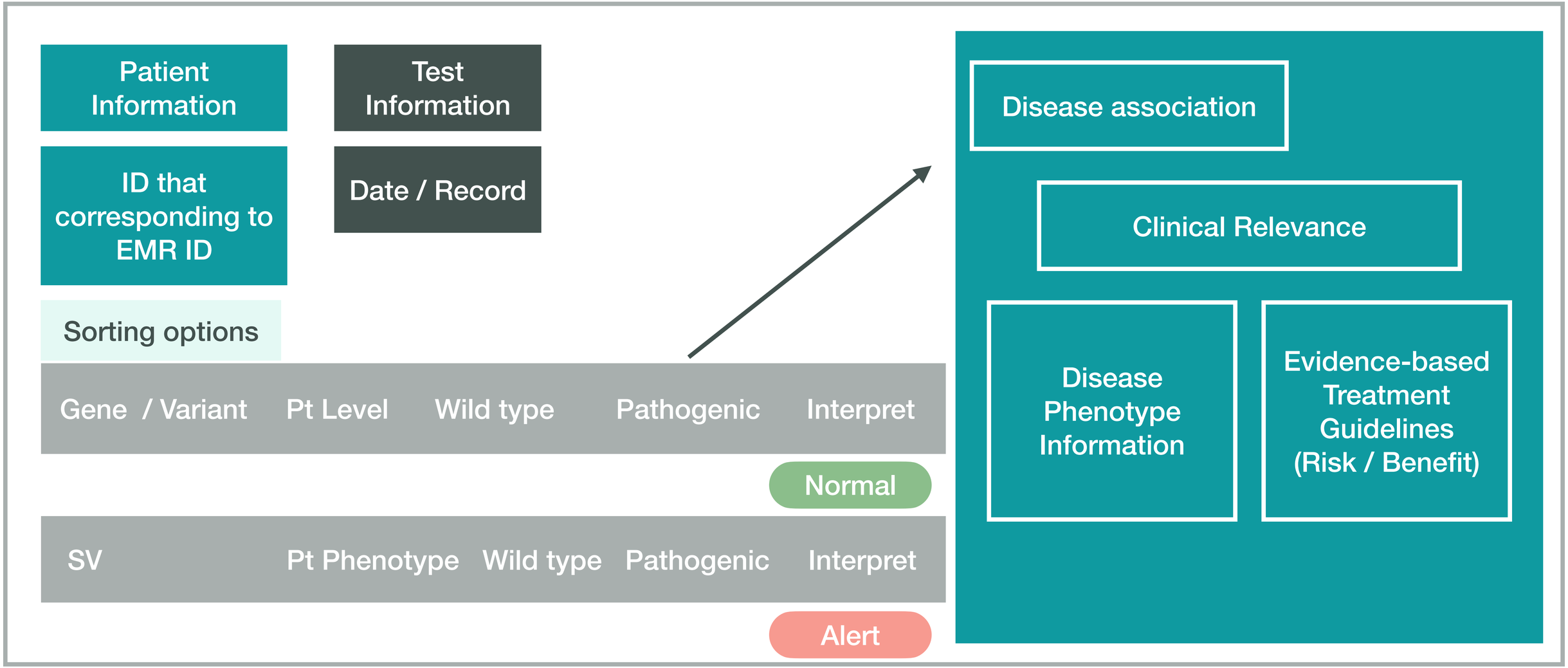
Non-genomic clinician Dashboard / Report

Information of interest: Genomics example

Normal

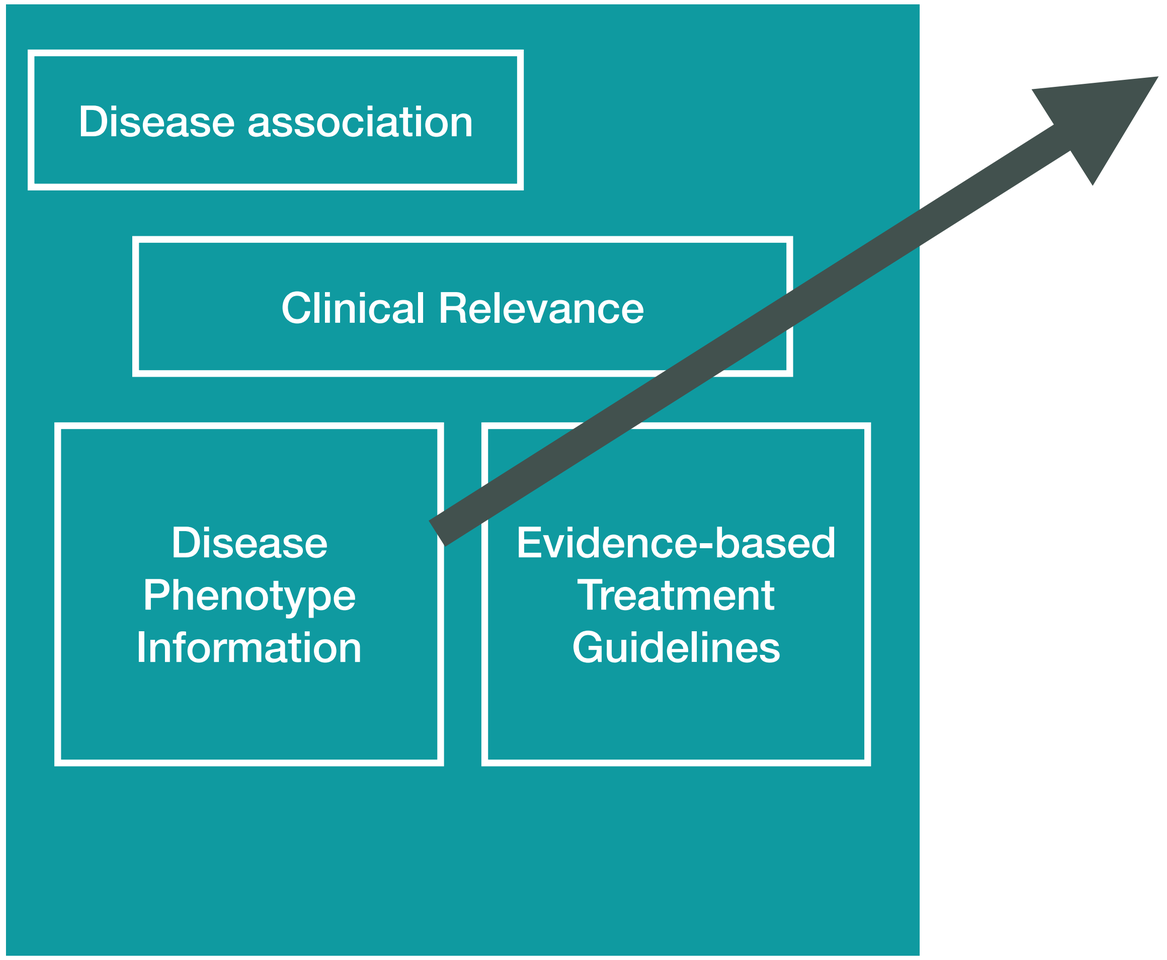
Abnormal

Alert



Non-genomic clinician Dashboard / Report

More detail on the information of clinical relevance

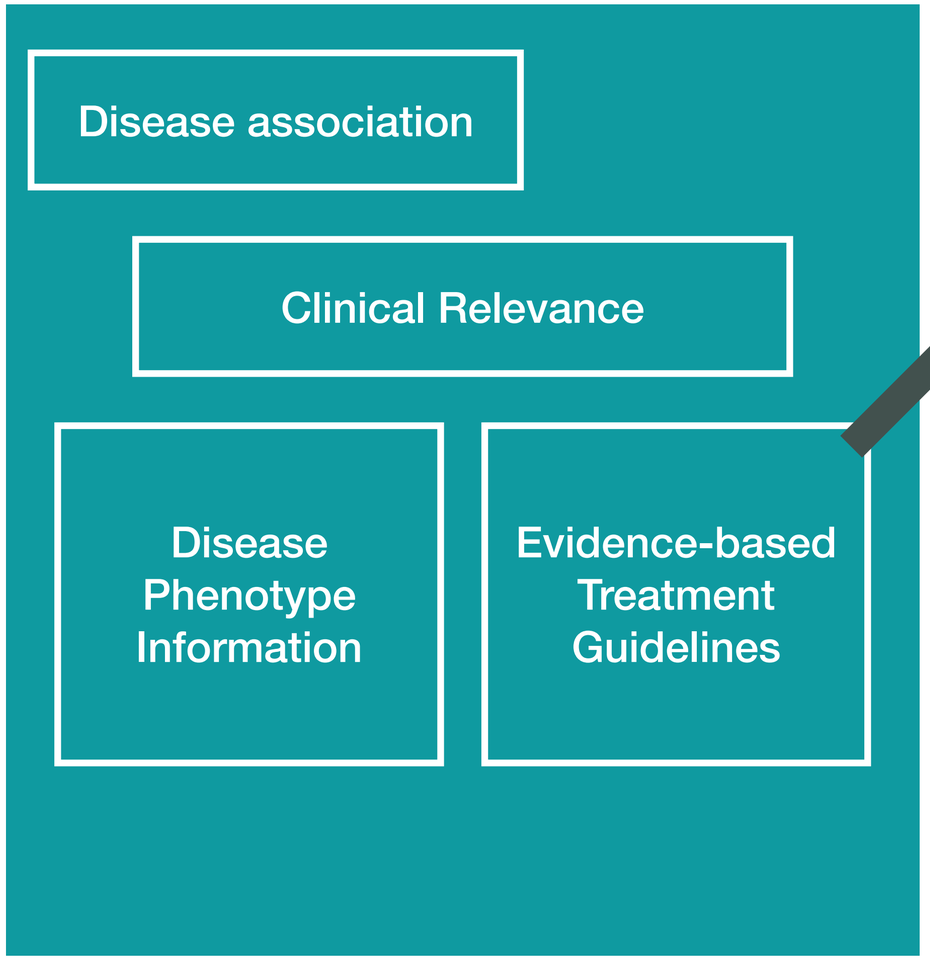


Disease Phenotype Information

- **Clinical Severity and Progression Course**
- **Symptoms**
- **Associated Lab abnormalities and other markers of differentiation**
- **Other unique aspects to this variant**
- **Previous interpretations of patient report**
- **If gene expression, historic trends in this patient**

Non-genomic clinician Dashboard / Report

More detail on the information of clinical relevance



Evidence-based Treatment Guidelines

- Clinical Risks
- Contraindications to certain treatments
  - Drug toxicities / sensitivities
  - Side effects to be aware of

- Guidelines on treatment
  - Efficacy data
  - Algorithms / Scores
  - Dosing

Non-genomic clinician Dashboard / Report

Clinician Preferences and Context for Consideration

Non-genomic clinicians are primarily interested in information that changes clinical management (aka treatment)

- If seeing a particular abnormal gene expression doesn't change the treatment (or there are no alternative treatments that will make a difference with a variant, the physician would probably rather not see the expression information.

Treatments are rarely decided solely base on the result of a single test

- Total patient history, presentation, clinical information, and patient preference is taken into account to decide on treatment

For complex integration of information, evidence-based algorithms and guidelines are helpful

- In a clinician's mind, the result of such algorithms, even with a numerical input, often group a patient into a 3 qualitative categories for making clinical decisions (ie. low, medium, high risks)

Patient Report Principles

Patients require ultimate control & portability over their report & information

Patients must be able to:

- Access their report information freely and be able to restrict access.
- Share access their report with any clinician they give permission to, regardless of any medical or insurance network
- In general, pdf's or static documents should be minimized or avoided as much as possible

Pdf output of report should contain:

- Basic Patient Information
- Information about the test conducted
- Basic phenotype result
- The clinical rationale for why the test was conducted

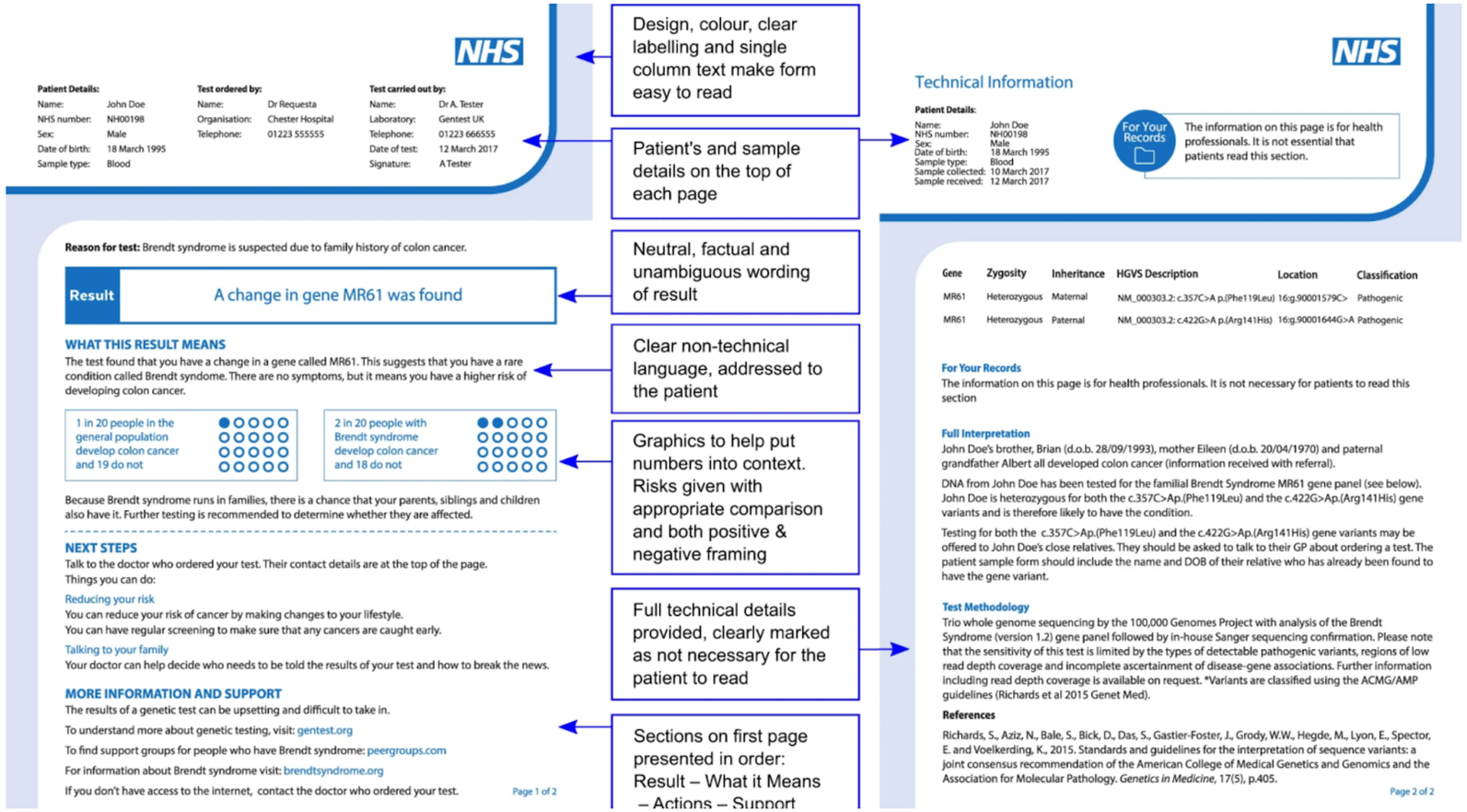
Interpretation of reports should be only available online

- Omics interpretation will continue to evolve and improve over time; using the latest interpretations is crucial to medical care
- Historical interpretations should be archived and accessible but not be materialized on static sheets such as pdf because it will unnecessarily burden medical providers to constantly chase down the most up-to-date report and interpretation for decision-making
- A QR code, as well as a typeable link to the online portal where the patient can access the latest interpretation of their report should be displayed on the report

Patient Report

Information of interest

Decent example to model (info content, not UI)



1. **Test result in simple terms**

2. **What the results mean**
(risk with appropriate visual comparison etc)

3. **Next Steps**
   - Followup with doctor to discuss
   - Reducing risk
   - Other support and resources

4. **Details (made clear not necessary to read)**
   - Full clinical interpretation
   - Test methodology
   - References

Patient Report

Pdf report

Patient Info

Date

Test Info

Phenotype result

Clinical rationale for test

Interpretation is online:

http://abcd.com/patientportal/324234452/login

Dashboard with interpretation

Patient Info

Date

Link to EMR notes

Phenotype Result

Clinical rational for test

Interpretation: What the results mean

Next Steps

Full details

Past interpretations archive