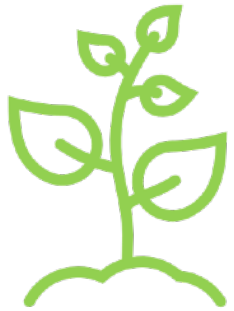
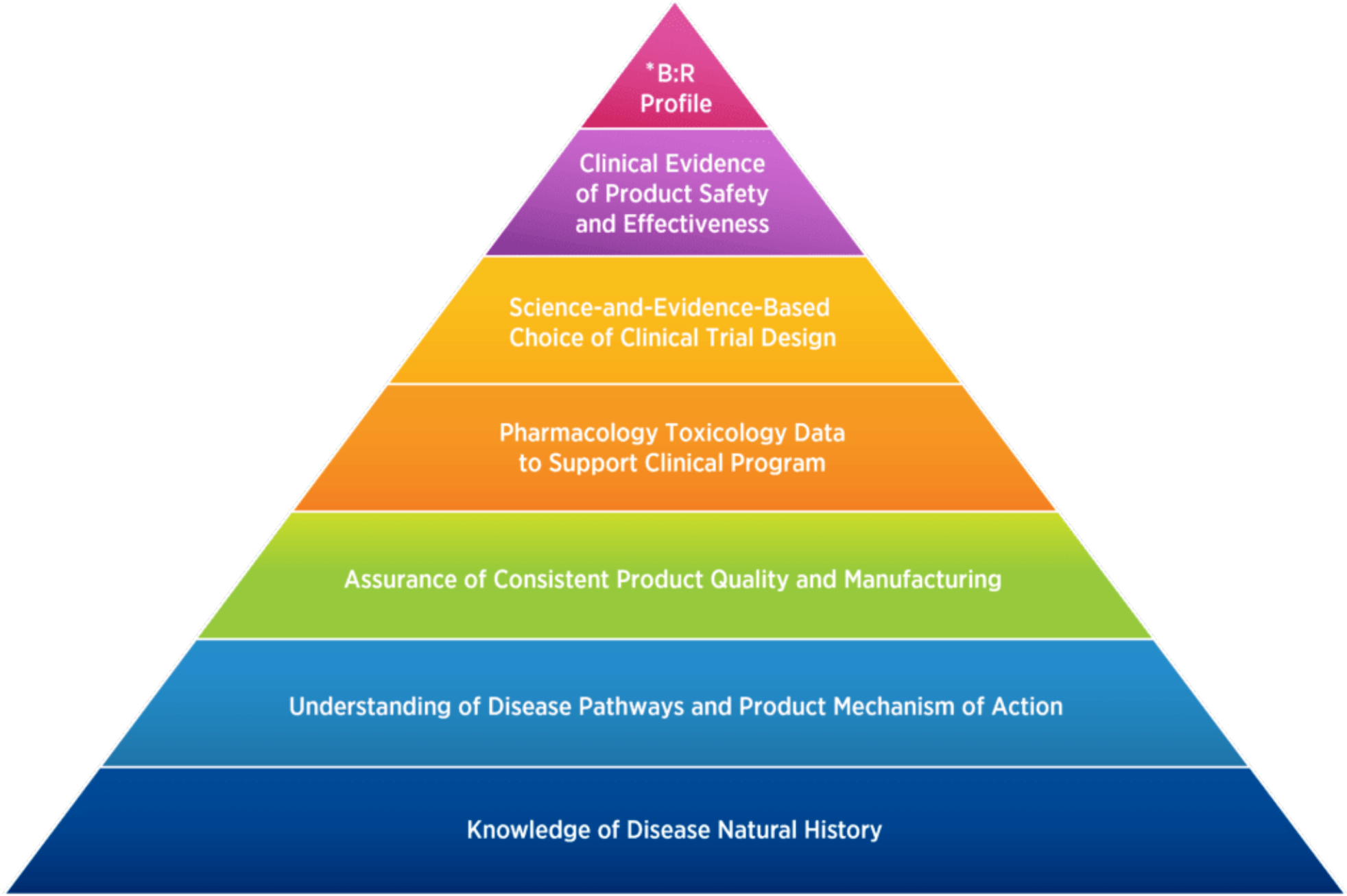
Natural history studies – the value of MaxHIGR

Indi Banerjee

Manchester UK





*B:R – Benefit : Risk

Determine Patients' Needs

Natural History Study

Natural history study databases contain more detailed clinical information over time, such as age at diagnosis, symptoms, medical images, and test results. Data may be entered by patients, their caregivers, or healthcare professionals.

- **Type of information** stored may include:
  - Patient contact information.

UK: Registry + Rare Disease BioResource

Public Health England

Protecting and improving the nation's health

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)

RaDaR Rare Diseases Registry Program

FROM MOLECULES TO MEDICINE: HOW PATIENTS CAN SHARE THEIR VOICES THROUGHOUT THE DRUG DEVELOPMENT PROCESS

WHERE PATIENTS CAN PARTICIPATE

Types of Participation	Basic Research	Preclinical Research	Clinical Research	Post-Marketing
Forming Patient Communities	✓	✓	✓	✓
Encouraging research	✓	✓	✓	✓
Funding research	✓	✓	✓	
Encouraging data sharing	✓	✓	✓	
Providing data	✓	✓	✓	✓
Participating in clinical trials	✓	✓	✓	
Participating in advisory committees	✓	✓	✓	✓
			✓	✓
				✓

HIGR  Max HIGR

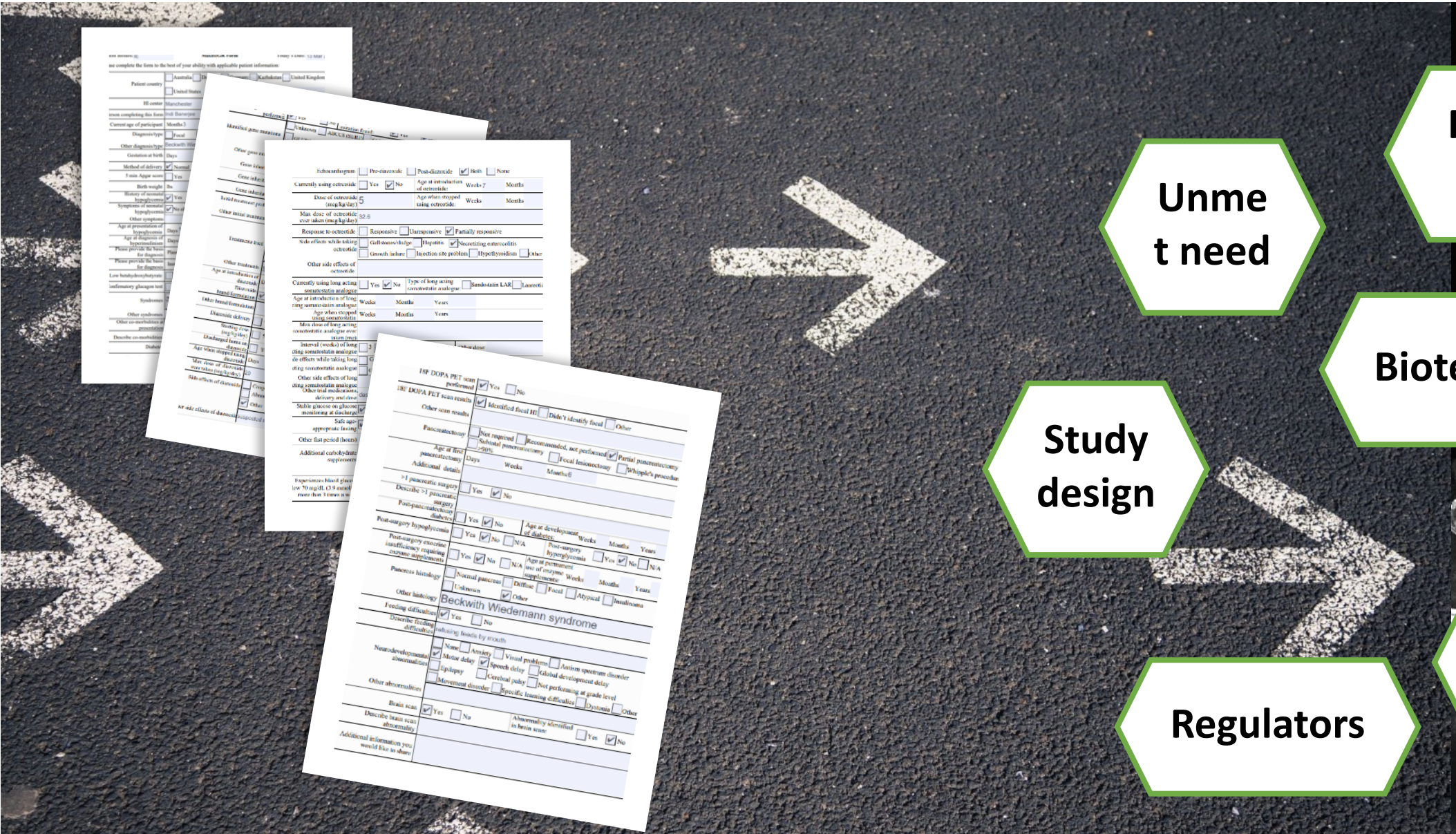
Patient Initials: IE

MaxHIGR Form

Today's Date: 13 Mar 23

Please complete the form to the best of your ability with applicable patient information:

Patient country	☐ Australia ☐ Denmark ☐ Germany ☐ Kazhakstan ☐ United Kingdom ☐ United States ☑ Other (please list): Ireland
HI center	Manchester
Person completing this form	Indi Banerjee
Current age of participant	Months 3 Years 2
Diagnosis/type	☐ Focal ☐ Diffuse ☐ Atypical ☑ Other
Other diagnosis/type	Beckwith Wiedemann syndrome
Gestation at birth	Days Weeks 39
Method of delivery	☑ Normal ☐ C-section ☐ Forceps
5 min Apgar score	☐ Yes ☑ No Apgar score 1-10: ?
Birth weight	lbs oz or gm 4500
History of neonatal hypoglycemia	☑ Yes ☐ No
Symptoms of neonatal hypoglycemia	☑ No obvious signs ☐ Excess jittery ☐ Seizures ☐ Unresponsive ☐ Other
Other symptoms	
Age at presentation of hypoglycemia	Days 1 Weeks Months
Age at diagnosis of hyperinsulinism	Days 11 Weeks Months
Please provide the basis for diagnosis	Plasma glucose: mg/dL or mmol/L 1.8
Please provide the basis for diagnosis	Insulin levels: pmol/L 23.6 or mU/L
Low betahydroxybutyrate	☐ Yes ☐ No Betahydroxybutyrate levels (mmol/L): ?
Confirmatory glucagon test	☐ Yes ☑ No
Syndromes	☑ Beckwith-Wiedemann ☐ Rubinstein Taybi ☐ Kabuki ☐ Turner ☐ Sotos ☐ Costello ☐ Fanconi ☐ Other
Other syndromes	
Other co-morbidities at presentation	☐ Yes ☑ No
Describe co-morbidities	
Diabetes	☐ Yes ☑ No
Describe diabetes	



Unmet need

Pharma

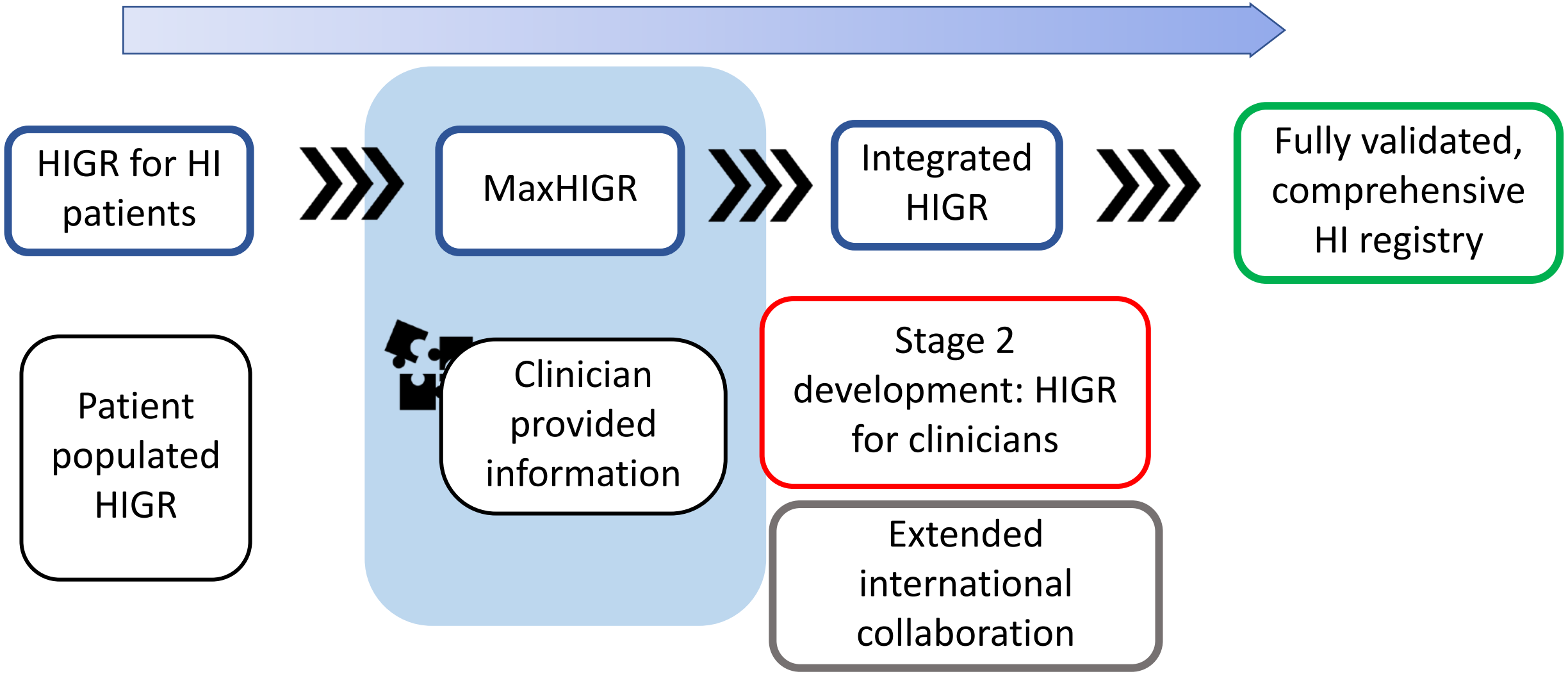
Biotech

Study design

Innovation

Regulators

Development of a comprehensive HI Global Registry



- Complete as much as you can
- Incomplete data is still valuable
- Your data entry is crucial to plan the next big step!