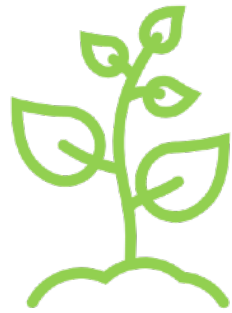
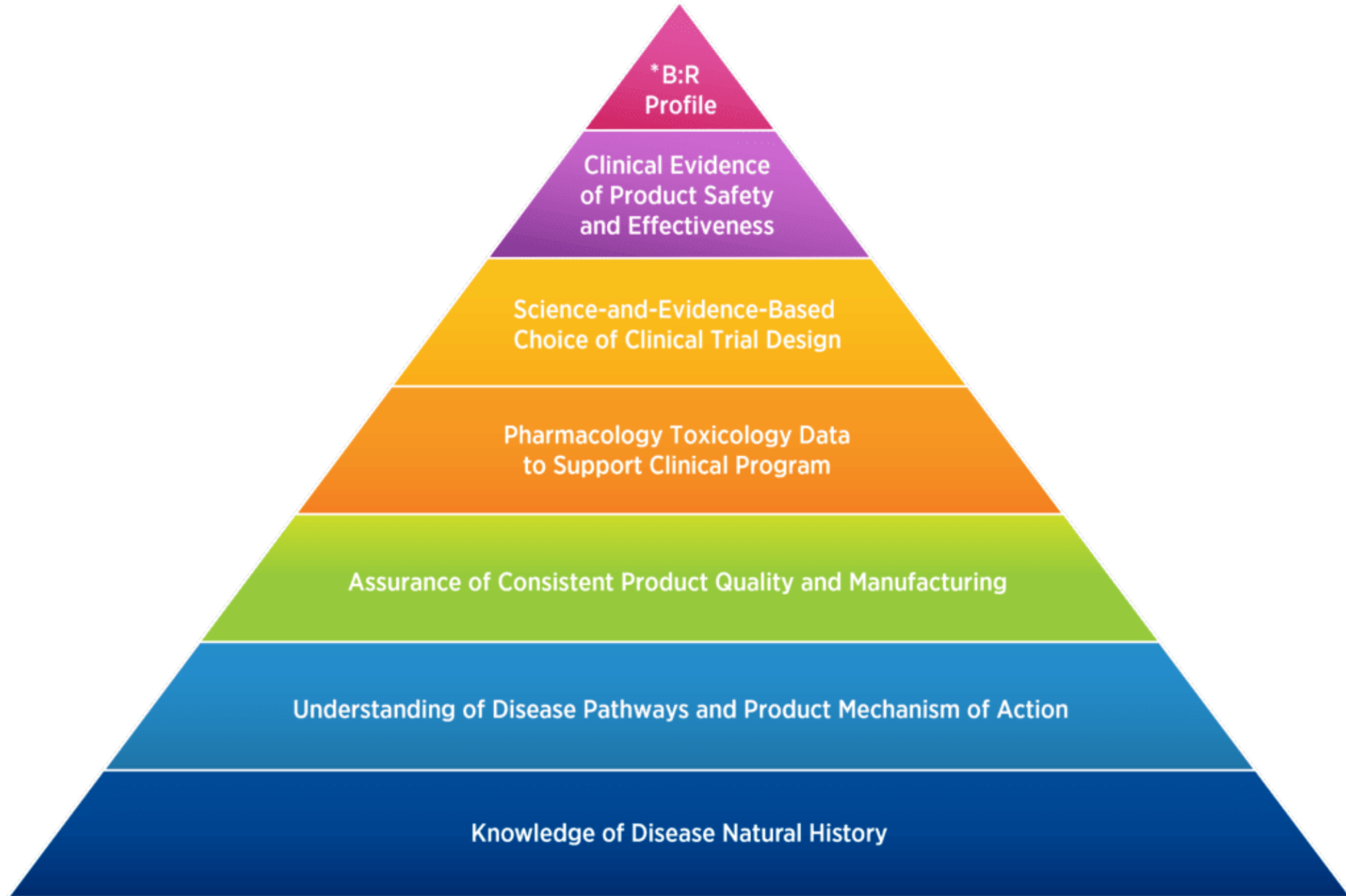


Natural history studies - the value of MaxHIGR



Indi Banerjee
Manchester UK





*B:R – Benefit : Risk



Determine Patients' Needs

OVERVIEW

PATIENT REGISTRIES

CONTACT REGISTRY

NATURAL HISTORY STUDY

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)

NIHR | BioResource

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Using our BioResource ▾

Centres & Programmes ▾

For recruiters ▾

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Rare Diseases BioResource

Around 1 in 17 people will develop a rare disease at some point in their lives. Today 400 million people in the world – and 3 million in the UK – have a rare disease. We support research into more than 60 disease areas, including immunology, neuroscience, haematology, rheumatology, cardiovascular disease, and many more.

**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	✓	✓	✓	✓
Advocating for			✓	✓
				✓

There Is Power in Numbers

HIGR



Max HIGR

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

Patient country	<input type="checkbox"/> Australia <input type="checkbox"/> Denmark <input type="checkbox"/> Germany <input type="checkbox"/> Kazhakstan <input type="checkbox"/> United Kingdom <input type="checkbox"/> United States <input checked="" type="checkbox"/> Other (please list): Ireland	
HI center	Manchester	
Person completing this form	Indi Banerjee	
Current age of participant	Months 3	Years 2
Diagnosis/type	<input type="checkbox"/> Focal <input type="checkbox"/> Diffuse <input type="checkbox"/> Atypical <input checked="" type="checkbox"/> Other	
Other diagnosis/type	Beckwith Wiedemann syndrome	
Gestation at birth	Days	Weeks 39
Method of delivery	<input checked="" type="checkbox"/> Normal <input type="checkbox"/> C-section <input type="checkbox"/> Forceps	
5 min Apgar score	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No	Apgar score 1-10: ?
Birth weight	lbs	oz or gm 4500
History of neonatal hypoglycemia	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No	
Symptoms of neonatal hypoglycemia	<input checked="" type="checkbox"/> No obvious signs <input type="checkbox"/> Excess jittery <input type="checkbox"/> Seizures <input type="checkbox"/> Unresponsive <input type="checkbox"/> 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	<input type="checkbox"/> Yes <input type="checkbox"/> No	Betahydroxybutyrate ₂ levels (mmol/L):
Confirmatory glucagon test	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No	
Syndromes	<input checked="" type="checkbox"/> Beckwith-Wiedemann <input type="checkbox"/> Rubinstein Taybi <input type="checkbox"/> Kabuki <input type="checkbox"/> Turner <input type="checkbox"/> Sotos <input type="checkbox"/> Costello <input type="checkbox"/> Fanconi <input type="checkbox"/> Other	
Other syndromes		
Other co-morbidities at presentation	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No	
Describe co-morbidities		
Diabetes	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No	
Describe diabetes		

PHARMACOPHYLACTIC FORM (FORM 1) (REVISED 12/2007)

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

Patient country: Australia Canada Kazakhstan United Kingdom United States

BE center: Manchester North Shoreline

Current age of participant: Months: Years:

Diagnosis type: Focal Multifocal

Other diagnosis type: Beckwith-Wiedemann syndrome

Duration at birth: Days:

Method of delivery: Normal Cesarean

5 min Apgar score: Yes No

Birth weight:

History of neonatal hypoglycemia: Yes No

Symptoms of neonatal hypoglycemia: Yes No

Age at presentation of hypoglycemia: Days:

Age at diagnosis of hypoglycemia: Days:

Please provide the name for diagnosis:

Please provide the name for diagnosis:

Low birthweight/erythrocytosis/inflammatory phagocyte test: Yes No

Syndromes: Yes No

Other conditions: Yes No

Other co-morbidities at presentation: Yes No

Diagnose co-morbidities:

Other side effects of diagnosis:

Education: Pre-diagnosis Post-diagnosis Both None

Currently using octreotide: Yes No

Age at introduction of octreotide: Weeks: Months:

Dose of octreotide (mcg/kg/day):

Age when stopped using octreotide: Weeks: Months:

Max dose of octreotide ever taken (mcg/kg/day):

Response to octreotide: Responsive Unresponsive Partially responsive

Side effects while taking octreotide: Gallstones/cholelithiasis Hypotonia Necrotizing enterocolitis Growth failure Infection at port site Hypothyroidism Other:

Other side effects of octreotide:

Currently using long acting somatostatin analogue: Yes No

Type of long acting somatostatin analogue: Sandostatin LAR Other:

Age at introduction of long acting somatostatin analogue: Weeks: Months: Years:

Age when stopped using somatostatin analogue: Weeks: Months: Years:

Max dose of long acting somatostatin analogue ever taken (mcg/kg/day):

Interval (weeks) of long acting somatostatin analogue:

Side effects while taking long acting somatostatin analogue:

Other side effects of long acting somatostatin analogue:

Other oral medications:

Subtle glucose on glucose monitoring at discharge: Yes No

Safe age appropriate feeding: Yes No

Other flat period (hours):

Additional carbohydrate supplements:

Expresses blood glucose < 70 mg/dL (3.9 mmol/L) more than 3 times in a row: Yes No

18F DOPA PET scan performed: Yes No

18F DOPA PET scan results: Identified focal HI Didn't identify focal Other

Other scan results: Not required Recommended, not performed Partial pancreatectomy Subtotal pancreatectomy Focal lesionectomy Whipple's procedure

Pancreatectomy: Yes No

Age at first pancreatectomy: Days: Weeks: Months: Years:

Additional details:

>1 pancreatectomy: Yes No

Describe >1 pancreatectomy:

Post-pancreatectomy diabetes: Yes No

Age at development of diabetes: Weeks: Months: Years:

Post-surgery hypoglycemia: Yes No

Post-surgery exocrine insufficiency requiring enzyme replacement: Yes No

Age at permanent hypoglycemia: Weeks: Months: Years:

Pancreas histology: Normal pancreas Diffuse Focal Atypical Insulomas

Other histology: Beckwith-Wiedemann syndrome Other:

Feeding difficulties: Yes No

Describe feeding difficulties:

Neurodevelopmental abnormalities: None Anxiety Visual problems Autism spectrum disorder Motor delay Speech delay Global development delay Epilepsy Cerebral palsy Movement disorder Specific learning difficulties Dyslexia Other:

Other abnormalities:

Brain scan: Yes No

Describe brain scan abnormality:

Abnormally identified in brain scan: Yes No

Additional information you would like to share:

Unmet need

Pharma

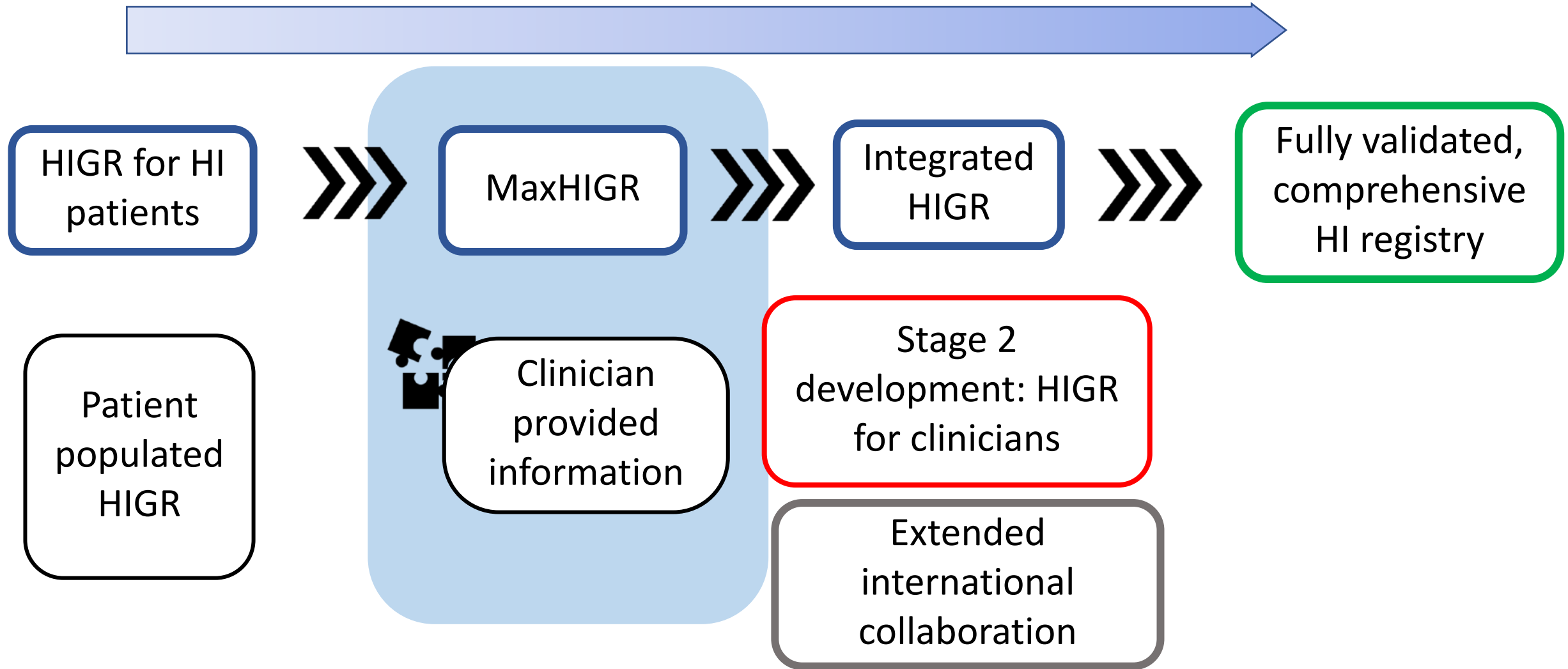
Biotech

Study design

Regulators

Innovation

Development of a comprehensive HI Global Registry



A background image of a bright blue sky with large, fluffy white cumulus clouds. The clouds are concentrated in the lower half of the frame, with the sky being a clear, vibrant blue above them.

- Complete as much as you can

- Incomplete data is still valuable

- Your data entry is crucial to plan the next big step!