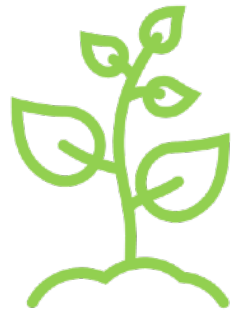
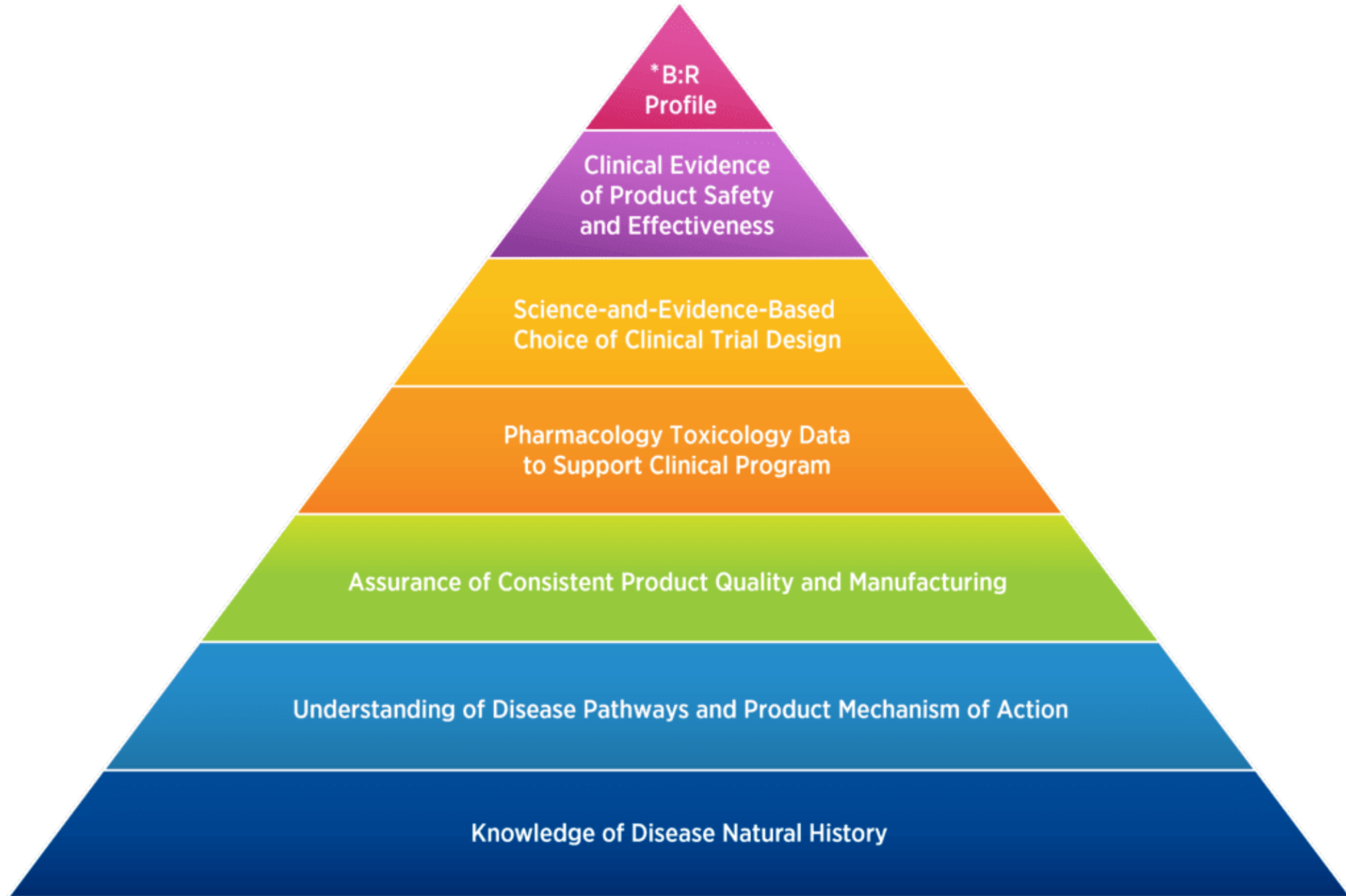


Natural history studies - from HIGR to MaxHIGR



Indi Banerjee
Manchester UK





*B:R – Benefit : Risk

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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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.



HI Global Registry (HIGR)

Comprehensive patient reported data

Clinician collected information
Electronic patient record
Local database
National Registry



Combined Medical + Parent
reported Registry

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 United States United Kingdom

BE center: Manchester Northampton

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

Significant neonatal hypoglycemia: Yes No

Other complications: None Other:

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: None Other:

Other conditions: None Other:

Other on-site findings at presentation: None Other:

Diagnosis confirmation: Disputed Other:

Age when stopped (weeks):

Age when stopped (months):

Age when stopped (years):

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

Interval (weeks) of long acting somatostatin analogue dose effects while taking long acting somatostatin analogue:

Other side effects of long acting somatostatin analogue:

Other side effects of short acting somatostatin analogue:

Subtle glucose on glucose monitoring at discharge: Yes No

Safe age appropriate fasting: Yes No

Other fast period (hours):

Additional carbohydrate supplements: Yes No

Experiences 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 resection Whipple's procedure

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

Additional details: >1 pancreatectomy Describe >1 pancreatectomy:

Post-pancreatectomy diabetes: Yes No

Post-surgery hypoglycemia: Yes No

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

Post-surgery exocrine insufficiency requiring enzyme replacement: Yes No

Post-surgery hypoglycemia: Yes No

Age at permanent hypoglycemia: Weeks: Months: Years:

Pancreas histology: Normal pancreas Diffuse Focal Atypical Insulomas

Other histology: Unknown Beckwith-Wiedemann syndrome

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: None Abnormally identified in brain scan Yes No

Brain scan: Yes No

Describe brain scan abnormality:

Additional information you would like to share:

Unmet need

Pharma

Biotech

Study design

Innovation

Regulators

Development of a comprehensive HI Global Registry

