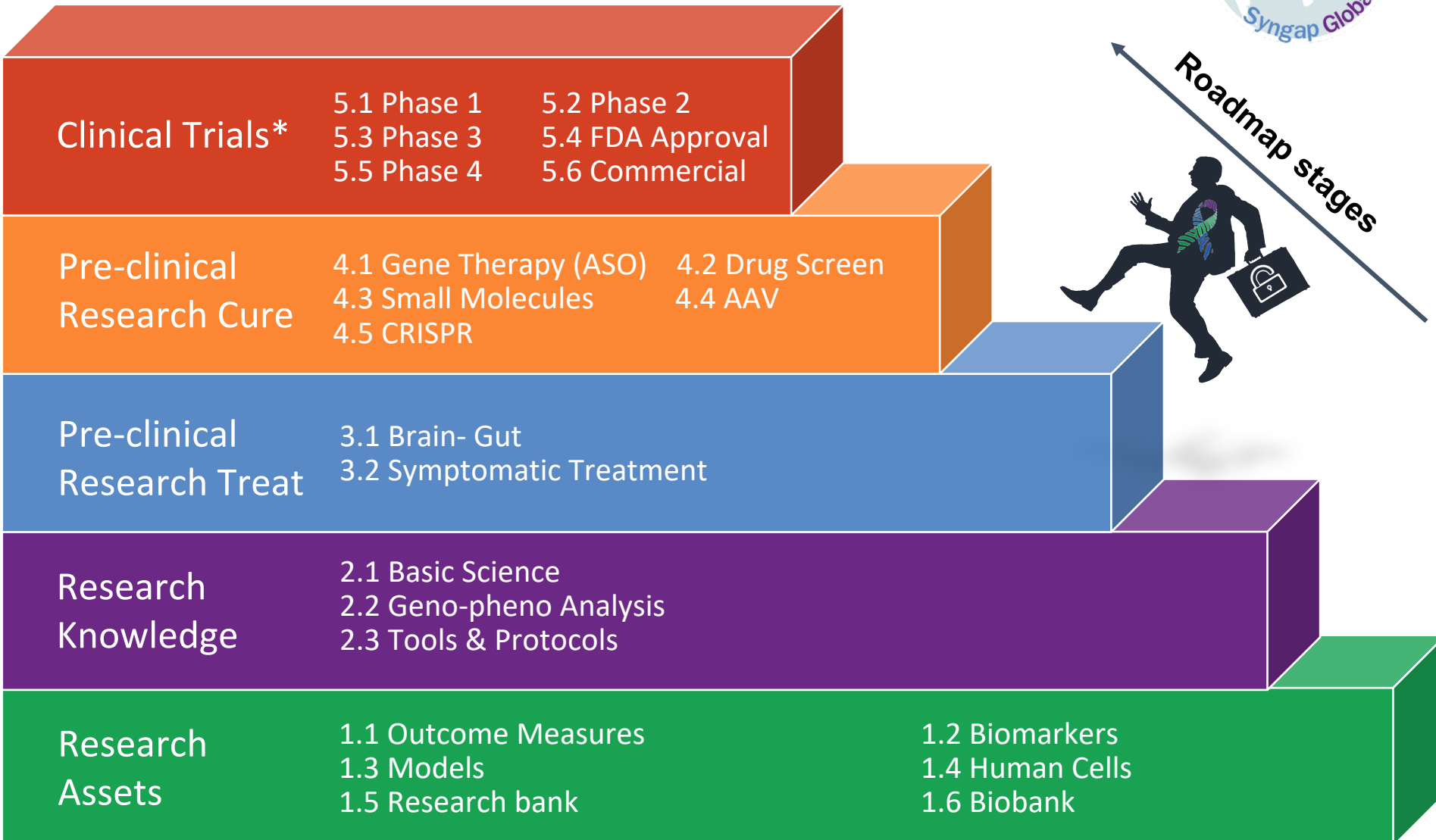


# The Syngap research roadmap stages




















## Why create a roadmap ?

- It's best practice to obtain a disease cure in the rare disease community
- Highlights future dependencies to ensure project timelines are aligned, eliminating potential bottlenecks
- Connect the community on projects
- Provides clear priorities for the community on paths to a cure and treatment
- Ensure our resources are aligned, focused, co-ordinated and not duplicated
- Establish Syngap community assets, making Syngap attractive for biotechs e.g. establish tools like a Natural history study, models



\* [Clinical trials phases based on the Dravet foundation clinical trial pipeline](https://www.dravetfoundation.org/dsf-funded-research/pipeline/)  
<https://www.dravetfoundation.org/dsf-funded-research/pipeline/>

# The Syngap research roadmap

Roadmap stage	Project	Lead Nation	Timeline			
			2020	H1 2021	H2 2021	2022+
Outcome measures	Natural History Study (Scheffer)	Australia 	◆			
Biomarkers	Biomarkers – gait analysis (Necker Hospital)	France 				
Models	Mouse models (Huganir, Grant, Komiyama). Multiple mutations (Brose, Holder, Rumbaugh)	Multiple 				
Human Cells	Patient Cell Lines ( Rumbaugh, Huganir, Clement, Petrou, Holder, Coba, Brüstle, Treutlein)	Multiple 	◆			
Basic Science	Animal based Syngap research C2 domain (Kind)	Austria 				
Basic Science	Molecular Syngap project (Sarah Shoichet)	Germany 				
Basic Science	Syngap protein synthesis – excitatory synapse (Clement)	India 				
Basic Science	Epigenetics project (Heller)	Multiple 				
Basic Science	SYNGAP isoform dynamics (Araki, Huganir)	US 				
Geno-phenol analysis	Effects on function of neurons comprising key striatal and dopaminergic circuits (Bateup)	US 				
Geno-phenol analysis	Geno-pheno analysis into hyperlocomotor activity, impaired memory, pain stimuli. (Nakajima, Miyakawa, Grant, Komiyama)	Japan 				
Geno-phenol analysis	EEG pathogenesis research (Kadam)	US 				
Symptomatic treatment	Microbiome study (Hsiao)	Australia 	◆			
Symptomatic treatment	Drug Screen (Rumbaugh, Clement), hiPSC (Bayes, Schülke)	Multiple 				
Symptomatic treatment	Statins (Kluger, Clement)	Multiple 				
Gene Therapy	3 projects; ASO (Petrou, Stoke), Natural ASO (Huganir)	Multiple 	◆			
Small molecules	Syngap regulator targets for small molecules, high throughput screen (Courtney)	Austria 	◆			
Small molecules	Testing pharmacological agents that target intracellular pathways (Ras, ERK, MTOR) to correct synaptic abnormalities (Michaud)	Canada 