

Overview

The TS Alliance supports initiatives to establish clinical resources for researchers in the pursuit of drug development and clinical care:

Natural History Database (NHD):

- Captures clinical data documenting the impact of TSC on the health of those affected over their lifetime
- 18 participating TSC clinics collect data, ensure quality in the data, and provide data distribution

Clinical Sites Across the US



TSC Clinical Sites

Clinical Site (Principal Investigator)	NHD # enrolled	BSR	RDCRN	PREVeNT
Minnesota Epilepsy Group, PA (Michael Frost, MD)	178			
Texas Scottish Rite Hospital for Children (Steven Sparagana, MD)	206			
NYU Langone Medical Center (Josiane LaJoie, MD)	93			
Herscot Center for TSC at MGH (Elizabeth Thiele, MD, PhD)	205			
Children's National Medical Center, DC (Wm. McClintock, MD)	111			
University of Chicago (Patti Ogden, APN FNP NP-C)	95			
UCSF Benioff Children's Hospital Oakland (Ali Mostajeleian, MD)	112			
UCLA (Joyce Wu, MD)	87			
University of Texas Health Science Center (Hope Northrup, MD)	111			
University of Alabama Birmingham (Martina Bebin, MD, MPA)	79			
The Cleveland Clinic (Ajay Gupta, MD)	91			
Children's Hospital Colorado (Susan Koh, MD)	114			
Nicklaus Children's Hospital Miami (Ian O'Neil Miller, MD)	79			
Loma Linda University Medical Center (Stephen Ashwal, MD)	82			
Univ. of Pennsylvania Medical Ctr. (Katherine Nathanson, MD)	67			
Boston Children's Hospital (Mustafa Sahin, MD, PhD)	85			
Cincinnati Children's Hospital (Darcy Krueger, MD, PhD)	309			
Washington University St. Louis (Michael Wong, MD, PhD)	81			
TS Alliance (Jo Anne Nakagawa)	37			
Duke University (Klaus Werner, MD, PhD)				
Children's Hospital Orange County (Mary Zupanc, MD)				
Children's Hospital Philadelphia (Katherine Taub, MD)				
Lucille Packard Children's Stanford (Brenda Porter, MD, PhD)				
Beaumont Children's Hospital (Danielle Nolan, MD)				

Research Category	Number of papers	Most recent publication using data from NHD
CNS and TAND	7	CNS: Song J, et al. Epilepsy treatment patterns among patients with tuberous sclerosis complex. <i>J Neurol Sci.</i> 2018;391:104-108. doi: 10.1016/j.jns.2018.06.011 .
Ophthalmology	1	The authors found that over 64% of individuals in the database who were prescribed drugs for epilepsy used three or more antiepileptic drugs. Over 22% had epilepsy surgery after trying antiepileptic drugs, and 35% had additional surgery after the first epilepsy surgery.
Urology	1	

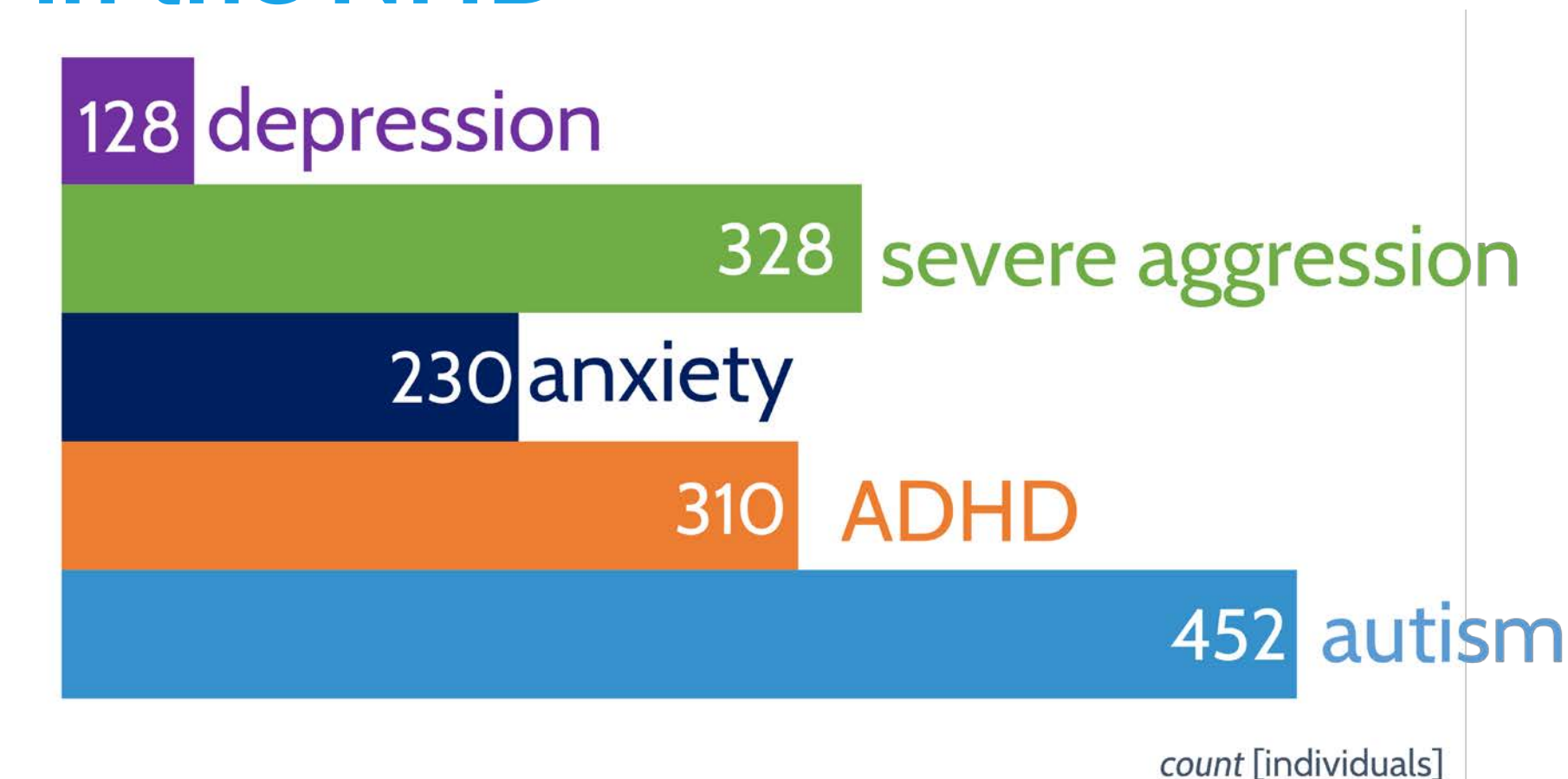
Biosample Repository (BSR):

- A biobank of tissues, blood, and DNA with corresponding patient clinical data
- Repository currently has 867 biosamples from 639 TSC patients and 166 biological parents
 - 579 linked to entries in the NHD
 - Additional 263 from the Rare Diseases Clinical Research Network (RDCRN) Synaptopathies Consortium and PREVeNT clinical trial

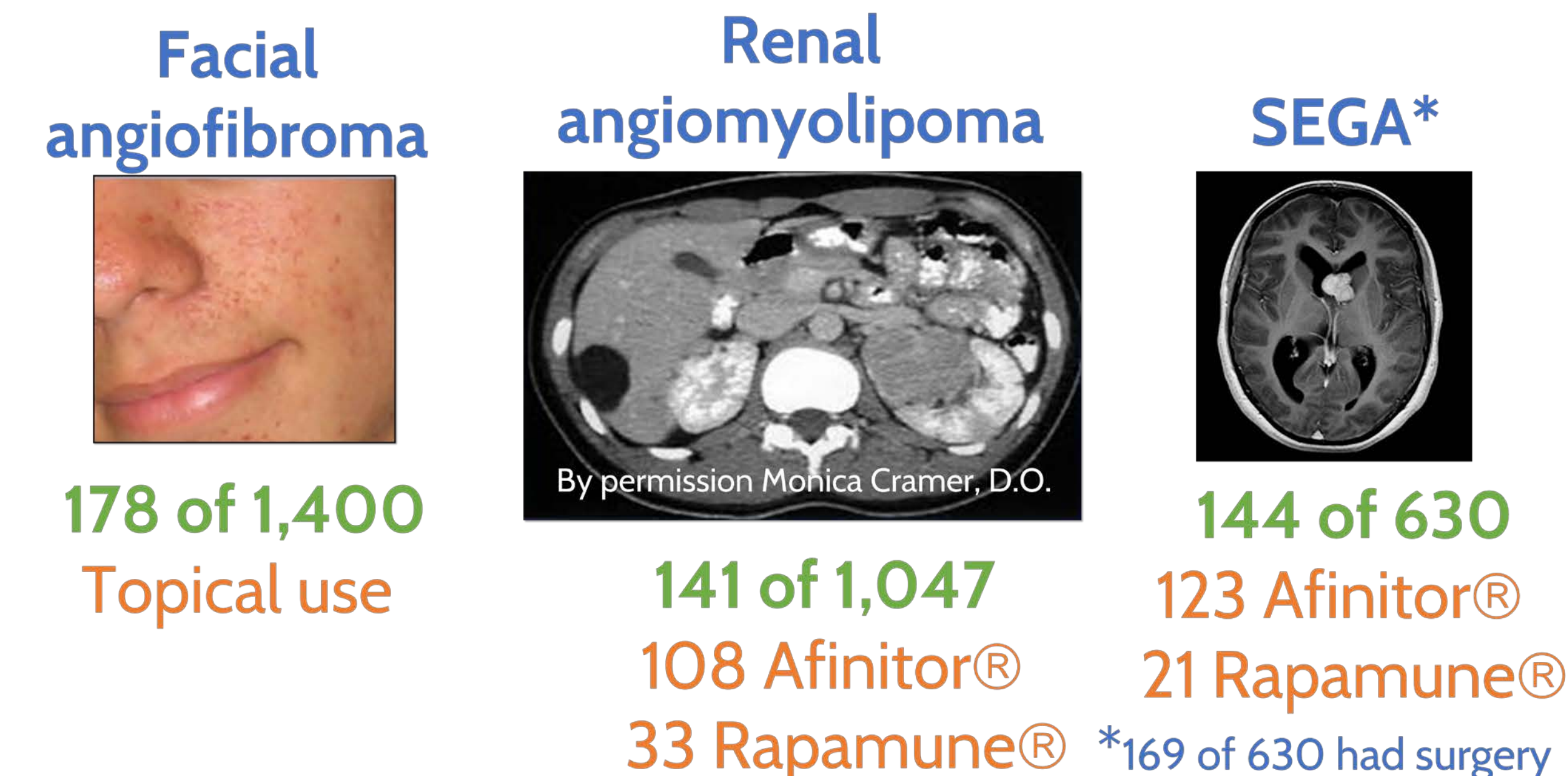
Footprint of Clinical Resources in the US

- 18 TSC clinics recognized by the TS Alliance participate in NHD and BSR
- We continually seek new participation for the collection of biosamples and clinical data

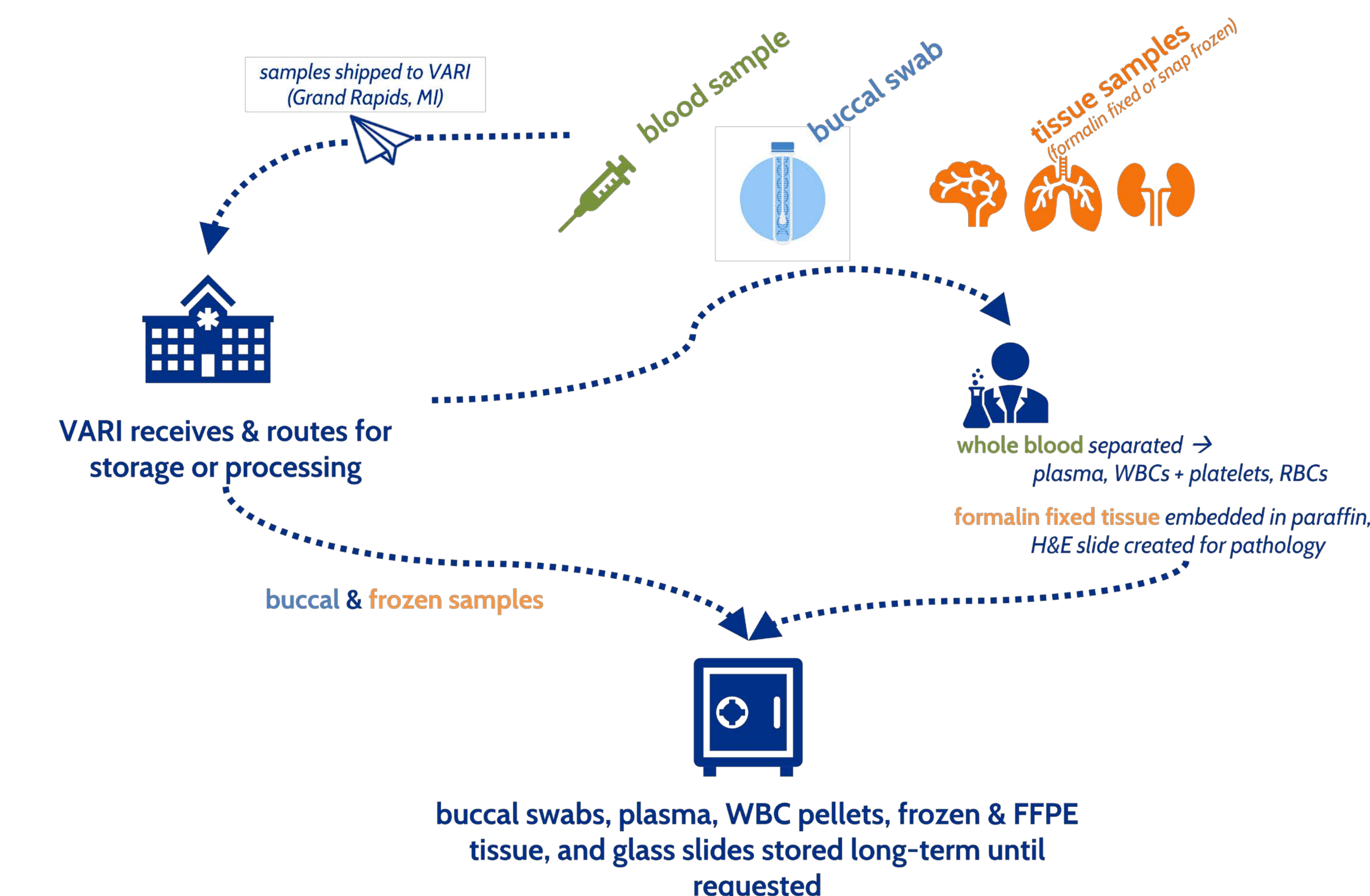
TSC Associated Neuropsychiatric Disorders (TAND) in the NHD



mTORi Use in Patients Enrolled in NHD



Van Andel Research Institute (VARI) Collaboration



Accessing Biosamples – Application Process

Researcher submits request for biospecimens



- ✓ Application for biosample access
- ✓ Investigator CV or biosketch
- ✓ IRB approval or exemption letter

Application reviewed by steering committee



- ✓ Sample availability
- ✓ Scientific merit
- ✓ Benefits of proposal

Request approved



- ✓ Requested modifications made (if necessary due to sample availability)
- ✓ Pi's institution completes MTA with TS alliance

Samples processed by VARI in accordance with request



- ✓ Plasma, WBC pellets, DNA, or RNA
- ✓ RNA paxgene fixed blood samples
- ✓ FFPE tissue sections, cores, or curls
- ✓ Frozen tissue sections, cores, or curls



Samples shipped to researcher!



For more information contact Jo Anne Nakagawa,
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