

Attendees: Subgroup leads: Isabella (Bella) Plumptre & Dawn Eichenfield
Members: Nika Finelt, Maria “Laura” Cossio, Maria Buethe, Camila Downey, Colleen Cotton, Joyce Teng, Aimee Smidt, Bernard Cohen, Jinia El-Feghaly, Cheryl Bayart, Elena Pope, Lara Wine Lee, Carolena Estee Steinberg

Staff: Katherine Devenport

Project Updates/Completions

Project 1: KHE cohort project

- o **Type of study:** Retrospective cohort
- o **Project Lead:** Elena Pope
- o **Recruitment help:** 71 patients enrolled, goal is 200. 12 sites involved, of which 3 are IRB approved and recruiting. They are recruiting patients and open to new sites. Legal team is working on contracts for every site.
- o **Timeline:** Hoping to complete recruitment by Dec 2024. Grant is for 2 years.
- o **Contact:** Elena Pope elena.pope@sickkids.ca

Project 2: Retrospective review of systemic therapy (MEK, PI3K, mTOR inhibitors) use for vascular anomalies

- o **Type of study:** Multicenter Review of Outcomes & Safety Profiles
- o **Project Lead:** Joyce Teng (with students Carolena & Jordan)
- o **Recruitment help:** Sites needed (18 sites currently, 10 have done IRB). Aiming for 200+ patients on any targeted therapy (especially sirolimus, trametinib and alpelisib).
- o **Timeline:** Open to enrolling new sites for next few months. Aiming to have IRB’s & DUA’s finalized by early summer, and complete data collection by end of summer.
- o **Contacts:** Carolena Estee Steinberg cesteinb@stanford.edu and Jordan Kim Uyen Bui jkbui@stanford.edu

Project 3: Vascular Anomalies Genotype-Phenotype Project (*not discussed today*)

- o **Project Lead:** Beth Drolet and Maya Muldowney
- o **Recruitment help:** Sites needed (12-14 sites), still enrolling patients (>500 patients enrolled)- in particular verrucous venous malformation cases and those with double hit mutations
- o **Contact:** Ellen Bruhn ebruhn@dermatology.wisc.edu
 - Two manuscripts anticipated– GNA11 R183 variants, capillary malformation genotype/phenotype correlation

Project 4: CM-AVM Syndrome Consensus Guidelines

- o **Project Lead:** Lara Wine Lee and Alexandra Richmond

- o **Updates:** Final round of surveys are in progress over the next few weeks, poster presentation at ISSVA in May, and currently working on manuscript.

New projects/ideas:

Project 1: Retrospective study of CM-AVM

- o **Type of study:** Retrospective cohort study, with possibility of prospective/registry study in the future.
 - Details include imaging, genotype/phenotype, and natural history
- o **Project Lead:** Lara Wine Lee
 - Team: Joyce Teng, Jinia El-Feghaly
- o **Update:** Looking to recruit additional leadership for this project to help with detailed study plan and commit to getting this project off the ground.
 - Especially those with prior registry experience. KHE registry study could be used as a template. Would be a long term project.
- o **Timeline:** Looking to start planning this project once consensus guidelines for CM-AVM are complete.
- o **Contact:** Lara Wine Lee winelee@musc.edu if interested in being a part of leadership working group for this project.

Project 2: Transitions of Care in vascular anomalies

- o **Type of study:** Survey of PEDRA members/vascular anomalies centers
- o **Project Lead:** Maria Buethe
 - Team: Joyce Teng, Bella Plumtre, Dawn Eichenfield
- o **Ideas:**
 - Some pedi hospitals won't see adults, but some can. For example, Joyce's vascular anomalies center is able to see adults, and they have e.g. pedi and adult IR specialists for head/neck and body respectively.
 - Key questions: How much of a problem is transition of care for vascular anomalies patients? What is the set up at different vascular anomalies centers? What adult services are offered? Are there age restrictions? What barriers exist? What could be some potential solutions?
 - PEDRA could send out study. Would require [PEDRA survey application form](#) which requires committee review (takes 6-8 weeks).
 - IRB: Colleen recommended we review requirements, as likely the main lead's institution IRB should host/approve IRB (or grant exemption).
 - Data collection: could be via redcap or qualtrics
- o **Timeline:** TBD
- o **Contact:** Maria Buethe mbuethe@health.ucsd.edu

Project 3: PTEN Management

- o **Project Lead:** Joyce Teng (with Carolina)
- o **Idea:** Looking at use of targeted therapies in patients with a PTEN mutation with a vascular anomaly.
 - May be sub project of their targeted therapy project above, given all patients who have PTEN mutation should be included on this
 - Considering PTEN patients on MEK inhibitor and sirolimus

- o **Timeline:** TBD
- o **Contact:** Carolena Estee Steinberg cesteinb@stanford.edu

Project 4: PG-like Growth in AVMs

- o **Project Lead:** Megha M. Tollefson
 - Not discussed in detail today
 - Bernard Cohen raised that he has had several patients with PG's associated with capillary malformations

Other Ideas:

- Bernard Cohen raised issue of no consensus for ulceration of infantile hemangiomas - will be discussed at main birthmarks session or next hemangioma subgroup meeting