

Elucidating the genetic basis of vein of Galen malformation

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Disclosures & Funding



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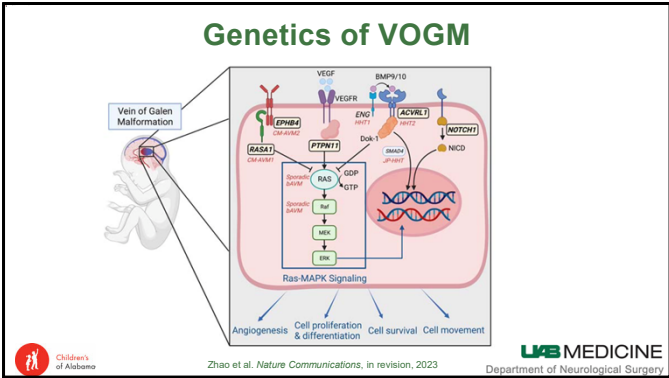


Challenges

- VOGM is a rare disease
- VOGM is thought develop during embryologic weeks 6-11
- Inability to sample somatic tissue as the lesion evolves over time



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Our central hypothesis is that inadequate access to VOGM tissue *in situ* hinders the identification of somatic mutations and transcriptional pathways responsible for VOGM pathogenesis and maintenance

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Approach

Endovascular Biopsy: Evaluating the Feasibility of Harvesting Endothelial Cells Using Detachable Coils

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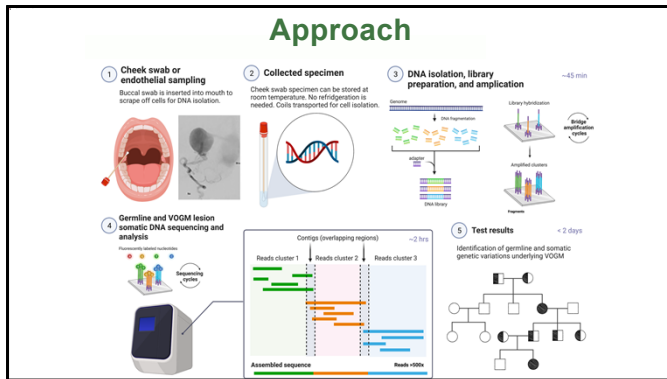
Endoluminal Biopsy for Molecular Profiling of Human Brain Vascular Malformations

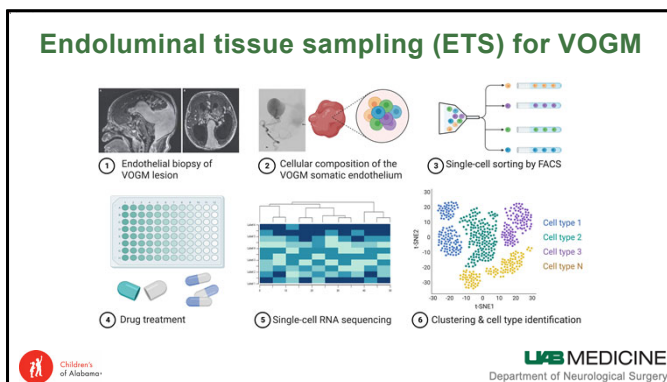
Elhan Winkler, MD, PhD, David Wu, BS, Eugene Gil, BS, David McCoy, MS, Kaitlin Harpeth, MD, Zhengda Sun, PhD, Kerstin Mueller, PhD, Jayden Rossi, BA, Helen Kim, PhD, Sharaz Wertschheimer, PhD, Mitchell Berger, MD, Torrance Nowakowski, PhD, Daniel Lim, MD, PhD, Aishwarya, MD, PhD, and Daniel Cooke, MD*
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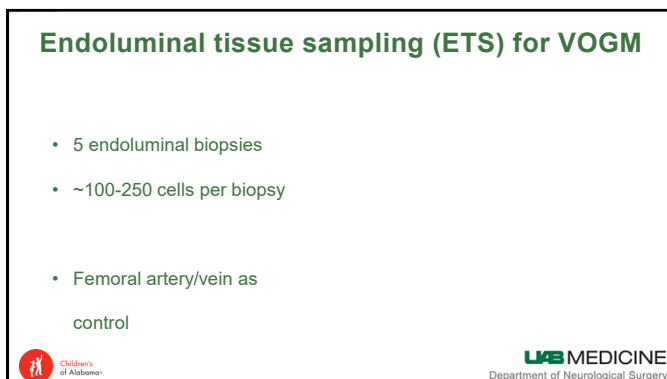
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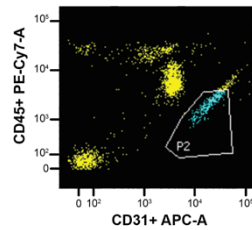






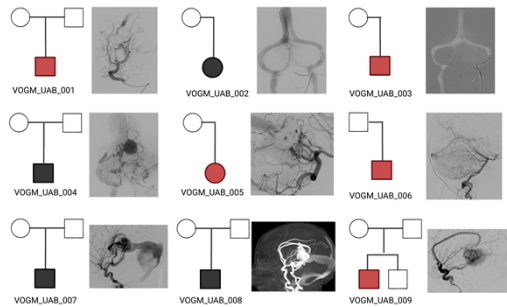
Endoluminal tissue sampling (ETS) for VOGM

- 5 endoluminal biopsies
- ~100-250 cells per biopsy
- Femoral artery/vein as control



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Pedigrees of VOGM families



*Red indicates endothelial sampling (ETS) performed



Vein of Galen Malformation

Genetics Research Consortium



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Ways to participate

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- 3) Endothelial sampling (ETS) of the VOGM lesion and collection of cheek swab DNA, sent to UAB/COA for analysis;
- 4) ETS of the VOGM lesion and collection of cheek swab DNA, analysis performed at individual site and shared with UAB/COA for meta-analysis



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Summary

- Approach to understand the genetic basis of VOGM



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- Approach to understand the genetic basis of VOGM
- ETS to understand somatic mutation and transcriptional pathways underlying VOGM



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- ETS to understand somatic mutation and transcriptional pathways underlying VOGM
- Creation of the VOGM-GRC



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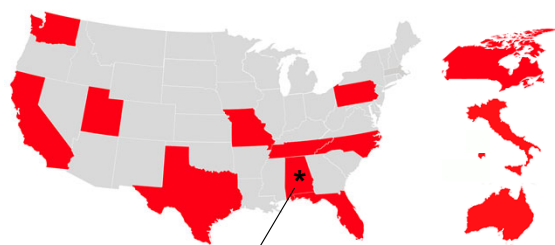
Summary

- Approach to understand the genetic basis of VOGM
- ETS to understand somatic mutation and transcriptional pathways underlying VOGM
- Creation of the VOGM-GRC
- Genetics may inform development of novel pharmacotherapeutics



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Beginnings of the VOGM-GRC...




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