Human Genetics Dispatch Summer 2023

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DOHG

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Chair's Corner

1 - Human Genetics Chair Peng Jin, PhD

Welcome to the summer edition of the Department of Human Genetics Dispatch for 2023.

Over the last several months, we find ourselves immersed in a blend of sorrow and joy. Our department is collectively grieving the loss of Cecelia Bellcross, a dedicated educator and researcher who was instrumental in establishing and directing our Genetics Counseling Training program for over a decade. Her energy, vision and genuine concern for both students and patients will be profoundly missed. We are grateful that Lauren Lichten, co-director of the program alongside Cecelia, has agreed to accept the role of Program Director and continue Cecelia's legacy.

On a more uplifting note, the future of biomedicine is brightly reflected in our Next Gen high school internship program. Now in its second year, the program has seen significant growth and progression. On July 6, our interns showcased their impressive video production and science communication skills in their presentations about their experiences. Emily Allen again did an amazing job, in both giving the interns a crash course in genetics and as convener of the program. Kudos also to Regina Gilbert and Aria Byrd for their considerable efforts in organizing food, transport, and other logistical matters for the interns.

Moreover, we are excited to announce that our inaugural class of Next Gen interns have made their university decisions. Out of the nine graduating seniors, three have chosen to continue their educational journey at Emory campuses, one has committed to Georgia Tech, while the others have secured spots at prestigious institutions such as Howard, Yale, Barnard, Brandeis, and Wake Forest. We eagerly anticipate their future collegiate achievements and the accomplishments that lie beyond.

Enjoy the rest of your summer!

Peng Jin, PhD

Professor and Chair

From our DEI Committee

DOHG events

A second round of DEI cohorts has begun. Cohorts are both a learning opportunity and a chance to get to know others in our Department. They are open to everyone: trainees, staff, and faculty. The time commitment is minimal (1 hour per month plus preparation time), but the impact is broad. This time around, there are three groups of about 5-6 meeting monthly until December. For questions, please email melanie.hardy@emory.edu.

In the fall, the department's **DEI Book Club** is planning to read Disability Visibility*: First-Person Stories from the Twenty-First Century,* edited by Alice Wong. Members held a ranked-choice vote on several proposed titles. In the meantime, we wish DEI co-chair Kate McCann all the best with her family expansion project!

Four Early Career/Young Investigator Seminars (conducted hybrid -- in person and with Zoom) are scheduled for this summer. Trainees can ask to have lunch with the speakers. All seminars are scheduled for **Fridays at noon**.

June 16 – Robert Fernandez, PhD, from Columbia, Talk title: "Dissecting the role of homeobox genes in the male C. elegans nervous system"

July 21 – Christopher Grochowski, PhD, from Baylor College of Medicine, Talk title: "Resolving complex genomic rearrangements"

August 4 – Thiago Arzua, PhD, from Columbia

August 18 – Monique Mendes, PhD, from Stanford

Emory events

University leaders are grappling with the recent Supreme Court decision limiting consideration of university applicants' racial/ethnic backgrounds. Interim SOM Dean Carlos del Rio has stated that within legal means, the School of Medicine will continue to foster a diverse community, both ensuring a richer learning experience and more equitable patient care and research.

Toolkit for international travelers facing identity-related challenges

Queer Woman of Emory lunch August 2, noon to 1:30PM at Miller Ward Alumni House

SOM community forum on engagement, equity and inclusion, August 17, noon to 1 pm, Register via Zoom

Department Spotlights

Get to know your DOHG colleagues. Each issue will feature randomly selected staff, faculty or trainee profiles.

*2 - **Professor Andrew Escayg, PhD***

Our neurogenetics lab works on epilepsy, neurodevelopmental disorders such as autism, and other mechanistically related disorders such as Alzheimer's disease. We utilize a wide range experimental approaches in order to 1) identify novel disease mutations, 2) better understand disease mechanisms, and 3) develop improved treatments. We are particularly interested in the generation of better treatments for severe forms of childhood epilepsy and treatment-resistant adult epilepsies.

I completed a B.S. in Chemistry and M.Phil. in Analytical Chemistry at the University of the West Indies, St. Augustine, Trinidad. I obtained my Ph.D. in Biochemistry from Lincoln University, New Zealand. I performed my postdoctoral research in the Department of Human Genetics, University of Michigan.

Outside of the lab, I enjoy running, working in the garden, and relaxing at the beach.

*3 - **Associate Scientist Teresa Douglas, PhD***

I am currently an Associate Scientist with Dr. Rani Singh's Metabolic Nutrition Program (MNP). My focus areas are translational research in the field of Inherited Metabolic Disorders (IMD), co-coordinating the eGNA Metabolic Nutrition ECHO training program, co-management of the MNP IMD -80°C Biospecimen Repository, planning and coordination of Metabolic Camp research activities, supportive grant writing and data management, along with mentoring undergraduate and graduate students on IMD relevant research projects.

My interest in IMD research and education was sparked by a lecture Dr. Singh gave in the nutrient metabolism course of Emory University's Nutrition and Health Science PhD program. Within the IMD field, I could see my interests in nutrition, biochemistry, and rare disease genetics woven together. Dr. Singh became my graduate research mentor, with an emphasis on phenylketonuria outcomes. I hope to expand my efforts towards rare disease advocacy, and researching nutrition relevant health outcomes in patients with MSUD and other rare genetic disorders.

Among the things I enjoy most is encouraging the next generation's interest in biomedical research and nutrition health. An example is the undergraduate Text to Table course that I co-instruct with Dr. Jill Welkley in the Center for the Study of Human Health, in collaboration with dietitian Carol Kelley and Emory Dining. For recreation, I enjoy dragon boating, spending quality

time with my nieces and nephews during the summer and with virtual reality, and going to local fairs and festivals.

*4 - **Graduate student Qile Dai***

I am currently a doctoral student in the Department of Biostatistics and Bioinformatics at Emory University, under the supervision of Dr. Michael Epstein and Dr. Jingjing Yang in the Human Genetics Department. My current research focuses on developing statistical and computational tools for two areas: (1) transcriptomic-wide association studies leveraging summary-level reference data, and (2) cell-cell communication inference using multi-subject single-cell RNA sequencing data.

Prior to joining Emory, I obtained my undergraduate degree in Economic Statistics from the University of International Business and Economics in 2018, and a master's degree in Biostatistics from Yale University in 2020. Outside of school, I enjoy cooking, playing board games, and listening to Korean pop music.

Key Discoveries: Human Genetics

How five ligand-receptor interactions together drive human astrocyte development

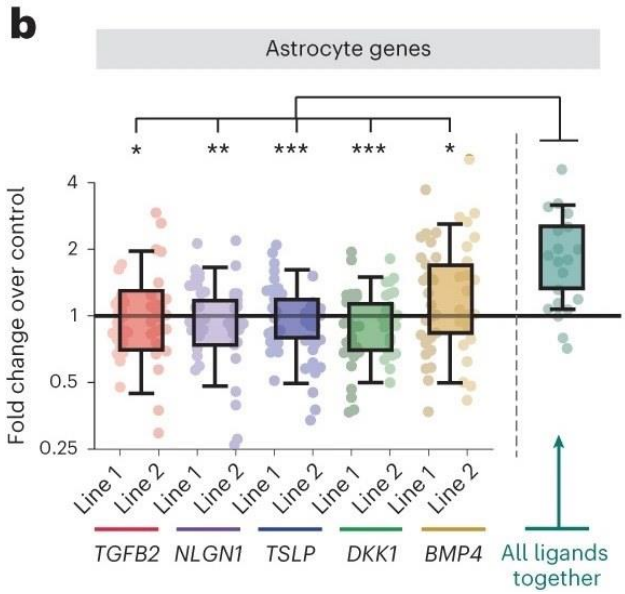
-- Steven Sloan, MD, PhD

Anna Voss*, Samantha Lanjewar and colleagues in the Sloan lab have gained new insights into how astrocytes are coaxed into being by their sibling cells. Astrocytes have been described as "the housekeepers and guardians of the central nervous system." They develop from the same progenitor cells as neurons, through a process called the gliogenic switch. The authors describe how five receptor-ligand interactions together drive this process.

As the authors report, the project began during COVID shutdown, when mining public gene expression datasets was a way to move forward despite restrictions on experimental work. Investigators' analysis using an algorithm called NicheNet led to some usual suspects (BMP4 and TGFb2) and some new candidates (NLGN1, TSLP, DKK1), whose combined effect on astrocyte development in culture was synergistic. They also explored downstream signaling pathways activated by these ligands. For more, check out Sloan's explanatory Twitter thread and *Nature Neuroscience*'s Research Briefing, which points to next steps -- such as assessing regional specificity and testing for the influence of other cell types.

*An undergraduate while in the Sloan lab, Voss is now a graduate student at Penn.

Nature Neuroscience, July 17, 2023, *Identification of ligand-receptor pairs that drive human astrocyte development*



5 - The five ligands' effect on astrocyte gene development is greater when combined.

Tracking locus coeruleus activity in rat Alzheimer's model

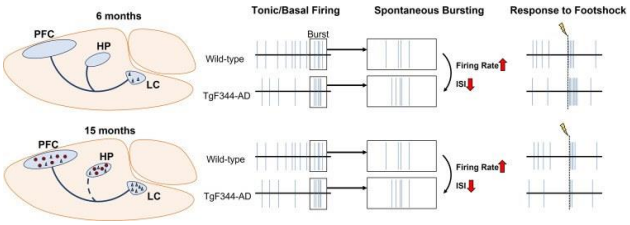
-- David Weinshenker, PhD

The locus coeruleus (LC), part of the brainstem, is a chief source for norepinephrine and an early site of neurodegeneration in Alzheimer's disease. It is also the site where early signs of Alzheimer's pathology appear in a transgenic rat model of AD called TgF344-AD. These animals express human APP (amyloid precursor protein) and human presenilin 1, both incorporating familial Alzheimer's mutations.

This paper, with graduate student Michael Kelberman as first author, focused on measuring locus coeruleus activity in these rats, both at baseline and in response to footshocks. It follows previous work from former postdoc Jacki Rorabaugh on the TgF344-AD rats' behavior and brain pathology. At 6 months of age, these rats display anxiety-like behavior, along with hyperphosphorylated tau protein in the LC. Later they develop deficits in learning and memory along with brain-wide tau and amyloid-beta.

At baseline, LC neurons from the transgenic rats were hypoactive compared with controls, but with elevated spontaneous bursting. Footshock responses depended on age, with hyperactivity evident at 6 months and hypoactivity at 15 months. These data further support the notion that early LC hyperactivity and late LC hypoactivity contribute to prodromal symptoms and cognitive/memory impairments of Alzheimer's.

Neurobiology of Aging, February 1, 2023, *Age-dependent dysregulation of locus coeruleus firing in a transgenic rat model of Alzheimer's disease*



6 - Summary of dysregulated LC firing patterns as a function of age in TgF344-AD rats

Novel tubulin chaperone variants linked to encephalopathy

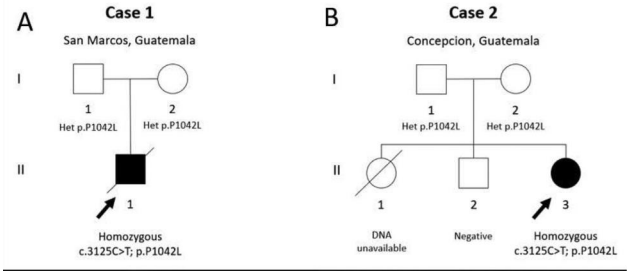
-- Juanita Neira, MD

Microtubules are present in all vertebrate cell types, but mutations disrupting them primarily impact neuronal function. This specific sensitivity is demonstrated in PEBAT (progressive encephalopathy and thin corpus callosum), which is highlighted in a recent publication by Juanita Neira and colleagues. The first author was Claudia Ocampo-Chih, a former Emory pediatrics resident who is now at Vanderbilt.

PEBAT – described as “an intriguing neurodegenerative tubulinopathy” -- is caused by mutations in TBCD (tubulin-specific chaperone D), which facilitates the polymerization and depolymerization of microtubules. In the paper, investigators describe two unrelated infants from Guatemala with PEBAT, hospitalized because of worsening neuromuscular weakness and respiratory failure. Both individuals were homozygous for a novel variant in TBCD: proline 1042 to leucine, which was identified through whole exome sequencing.

Working with Bo Liang and colleagues in the Department of Biochemistry, the authors visualized the effect of the mutation on the TBCD protein, concluding that the mutation is likely to disrupt protein structure and interactions. The authors also highlight the gap between reports of VUS (variants of unknown significance) and the resources available to investigate and reclassify such variants.

Pediatric Neurology, November 23, 2022, *PEBAT, an Intriguing Neurodegenerative Tubulinopathy Caused by a Novel Homozygous Variant in TBCD*



7 - PEBAT pedigrees from Ocampo-Chih et al (2023)

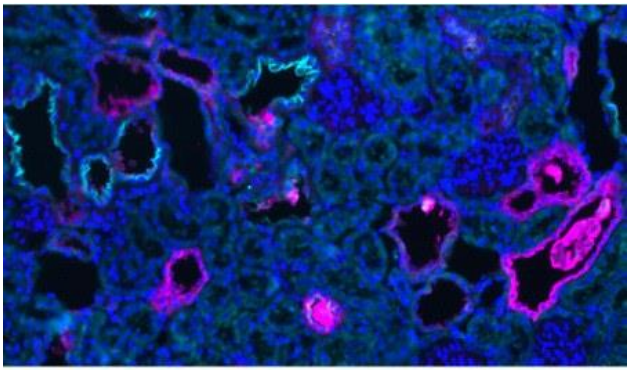
Ciliary role in kidney cyst development revealed

-- Tamary Caspary, PhD

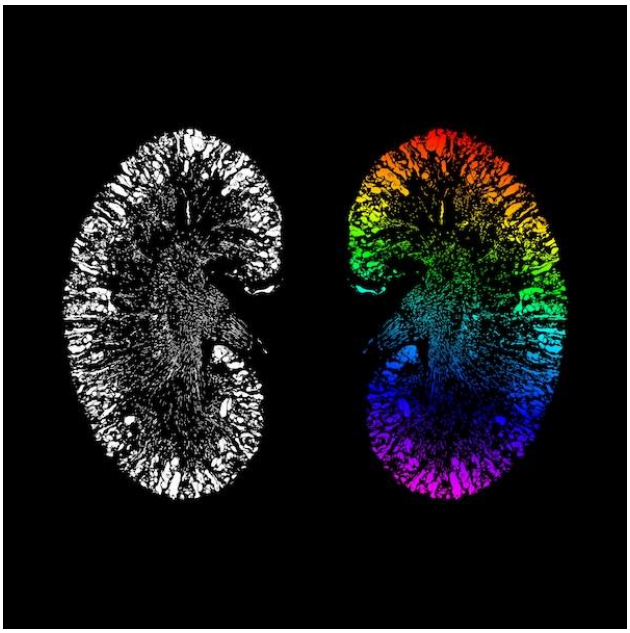
The Caspary lab has been using the GTPase ARL13B as a lens for understanding the critical roles of primary cilia across biology. Their recent work may provide insights into mechanisms of polycystic kidney disease, one of the most common genetic disorders.

As shown by the Caspary lab and others, complete deletion of ARL13B in kidneys results in cyst formation. In this paper, Robert Van Sciver and colleagues show that ARL13B's function preventing kidney cysts takes place within cilia. They do so by examining kidney development in mice with a knock-in form of ARL13B that is enzymatically active but not localized to cilia. These mice still develop kidney cysts. The authors also show that ARL13B's GEF (guanine nucleotide exchange factor) activity is not required to prevent kidney cysts.

Ciliary ARL13B inhibits developmental kidney cystogenesis in mouse, May 18, 2023, *Developmental Biology*



8 - Magenta indicates antibody to Tamm-Horsfall protein (THP)/uromodulin, revealing cysts in the thick ascending limb of the loop of Henle in Arl13b-V358A/V358A *kidneys. From van Sciver et al. (2023)*



9 - Cover image from Developmental Biology

From root to stem: transcriptome of developing human spinal cord

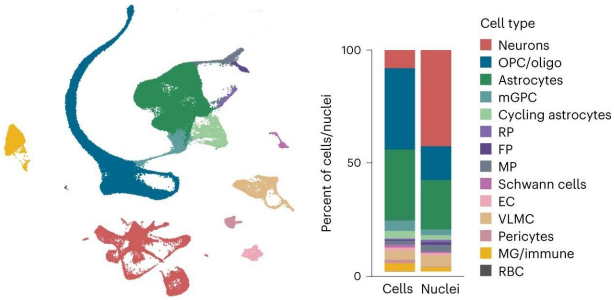
-- Jimena Andersen, PhD

Andersen and colleagues from the Pasca lab at Stanford have compiled transcriptome data from the developing human spinal cord. The authors profiled both cellular and nuclear RNA from spinal cord samples obtained from gestational week 17 and 18, using the 10x Genomics Chromium system to encapsulate single cells and sequence the RNA. They describe extensive transcriptome heterogeneity across and within cell types, both in terms of anatomical locations and white/gray matter.

The cell types covered include: neurons, oligodendrocytes, astrocytes, multipotent glia-derived progenitor cells, and microglia, along with pericytes, Schwann cells, endothelial cells, and vascular leptomeningeal cells. Within the category of neurons, the investigators separate glutamatergic (excitatory), GABAergic (inhibitory), and cholinergic neurons, and both visceral and skeletal motor neurons.

The authors anticipate that this resource, integrated with two other developing spinal cord data sets published in 2021, will contribute to future investigations of motor neuron diseases such as ALS, leukodystrophies, and Charcot-Marie-Tooth. The results are available online at http://devspinalcord.su.domains.

Single-cell transcriptomic landscape of the developing human spinal cord, April 24, 2023, *Nature Neuroscience*



10 - Representation of cell types covered by spinal cord transcriptome study

OTTERS: framework for TWAS analysis with larger datasets

-- Michael Epstein, PhD, Jingjing Yang, PhD

TWAS (transcriptome-wide association study) is an increasingly popular and powerful strategy for identifying genes that influence complex traits and risk for diseases such as Alzheimer's, cardiovascular disease and various cancers.

The Epstein and Yang labs have developed a framework for TWAS analysis that can make use of emerging summary-level eQTL (expression quantitative trait loci) results now being generated, which are derived from higher reference sample sizes -- sometimes 100 times more -- than previous studies.

The authors apply their method to results from the eQTLGen consortium and cardiovascular disease data from the UK Biobank, noting that their framework outperforms the popular FUSION approach. The authors note that the tool could also be applied to other QTL data types such as splicing, methylation, metabolomics or proteomics. The first author is graduate student Qile Dai (highlighted above in this newsletter).

Nature Communications, March 7, 2023, *OTTERS: a powerful TWAS framework leveraging summary-level reference data*

11 - OTTERS stands for: Omnibus Transcriptome Test using Expression Reference Summary data

Parent-of-origin effects: detective on the case

-- Michael Epstein, PhD

For some genetic variants, a “parent-of-origin effect” (POE) modulates the effect of an allele on a trait based on whether the allele came from the mother or father. While intrauterine and mitochondrial mechanisms can generate POEs, genomic imprinting is a major mechanism, with Prader-Willi and Angelman syndrome as classic examples of POE-associated diseases.

Since most GWAS studies sample unrelated individuals and parental relationships are unknown, sophisticated statistical methods are required to detect POE variants. Graduate student S. Taylor Head reports a method for improving discovery of POE variants, named after Agatha Christie’s detective: POIROT. The authors have applied POIROT to GWAS data from the UK Biobank using BMI and cholesterol phenotypes, and they identified 338 genome-wide significant loci for follow-up investigation.

Bioinformatics, April 17, 2023, *POIROT: a powerful test for parent-of-origin effects in unrelated samples leveraging multiple phenotypes*

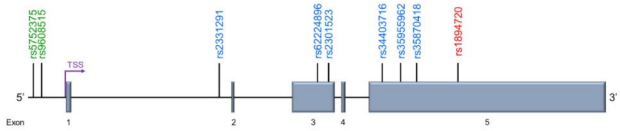
12 - POIROT aids in analyzing parent-of-origin effects

GOMAFU: RNA regulator of neural interferon response

-- Bing Yao, PhD, Jue Feng, PhD

The long non-coding RNA GOMAFU is dysregulated in brain tissue samples from people with schizophrenia and harbors variants that contribute to schizophrenia risk. Bing Yao's lab, together with Yue Feng's lab from Pharmacology and Chemical Biology, analyzed the function of GOMAFU in a neural progenitor cell model and in publicly available datasets. The authors report that GOMAFU is a suppressor of interferon responses, leading to hyperactive interferon pathways. The findings shed light on how immune disturbances in early life may increase risk for neuropsychiatric diseases. Assistant scientist Peng Teng and postdoctoral fellow Yangping Li are co-first authors.

Brain, Behavior and Immunity, June 8, 2023, *The human lncRNA GOMAFU suppresses neuronal interferon response pathways affected in neuropsychiatric diseases*



13 - SNPs in the GOMAFU *locus have been associated with acute myocardial infarction (Green), myocardial infarction (Blue), and schizophrenia (Red)*

Aggression and resistance: genome-wide analysis of social defeat stress

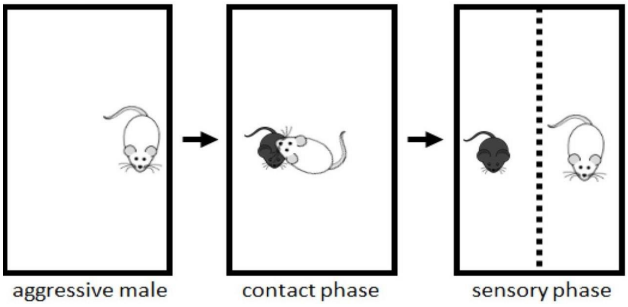
-- Bing Yao, PhD

The stress of chronic social defeat – exposure to an aggressor followed by sensory reminders of the encounter – generally induces depression- and anxiety-like behaviors in mice. Yet a minority of animals in a cohort display resistance to the effects of this stress protocol, providing

investigators an opportunity to probe gene expression and DNA modification patterns, in relation to stress responses and resistance.

In their publication, Janise Kuehner and colleagues in Bing Yao's lab analyze genome-wide patterns of social defeat stress on DNA methylation and hydroxymethylation. Their study is the first to simultaneously profile paired 5mC, 5hmC, and transcriptome data across three different timeframes of social defeat (acute, chronic and longitudinal). The authors detect evidence for epigenetic memory protecting against chronic stress, and highlight the involvement of immune and metabolic pathways.

G3 (Genes, Genomes, Genetics), May 25, 2023, *Social defeat stress induces genome-wide 5mC and 5hmC alterations in the mouse brain*



Racial/ethnic diversity in classic and clinical variant galactosemia

-- Judy Fridovich-Keil, PhD

The families affected by classic galactosemia in the United States are racially and ethnically diverse, mirroring the population as a whole. Yet previous studies of long-term outcomes in galactosemia have relied upon a predominantly White/Caucasian patient pool. Working with Judy Fridovich-Keil and David Cutler, undergraduate student Nichole Stettner (see photo) highlighted this gap by analyzing galactosemia prevalence among various groups, as reported by state newborn screening programs.

Molecular Genetics and Metabolism, February 21, 2023, *Racial and ethnic diversity of classic and clinical variant galactosemia in the United States*

14 - In May, Nichole Stettner completed her Emory undergraduate degree summa cum laude.

Telehealth and genetic counseling: implications for student supervision

-- Lauren Lichten, MS, CGC, Cecelia Bellcross, PhD

For genetic counselors, telehealth was a critical mode of delivery during the COVID-19 pandemic, and it is anticipated to be an important part of future healthcare delivery and education as well. A survey conducted by Lauren Lichten, Cecelia Bellcross and colleagues found that genetic counselors prefer in-person service delivery for student supervision, even more than for patient care. At the same time, genetic counselors will need to be prepared to provide telehealth counseling services and student supervision in the future, regardless of their preferences, the authors advise.

The authors compiled 132 survey responses from GCs in the United States and Canada. According to PubMed, this is the only paper that Lichten and the late Bellcross authored together. It was part of Lichten's Woodruff Health Educators Academy Fellowship, supported through the Audrey Heimler Special Project Award from the National Society of Genetic Counselors.

Journal of Genetic Counseling, April 24, 2023, Genetic counselors' perceptions of student supervision across service delivery models.

15 - The Heimler Special Project Award was aimed at developing a guide for telehealth student supervision in genetic counseling.

Probing Polycomb function in lung cancer and COPD

-- Aria Byrd, PhD

Here we highlight two publications from Aria Byrd, coming from her work in Christine Fillmore Brainson's lab at University of Kentucky. Byrd is assistant director of the Warren National Fragile X Research Center and assistant director of the department's NextGen outreach program.

The Polycomb group genes were first defined in *Drosophila* as regulators of homeobox gene expression. Biochemical studies have revealed the histone methyltransferase EZH2 as an enzymatic component of the Polycomb complex, which promotes a repressive chromatin state. In 2020, an EZH2 inhibitor was approved for certain cancer types, and clinical studies have been exploring how EZH2 inhibitors can be combined with other drugs.

In a *Nature Communications* paper, Byrd and her colleagues studied the effects of deleting EZH2 in K-ras-driven lung cancer cells. They found vulnerabilities for EZH2-deficient tumors, pointing toward potential drug combinations. In *Stem Cell Reports*, Byrd and her team showed how reduction of Polycomb/EZH2 activity could play a role in chronic obstructive pulmonary disease. In lung organoids, the authors demonstrated that EZH2 loss leads to reduced self-renewal capacity and a skewing of development towards squamous cells.

Nature Communications, January 20, 2023, Polycomb deficiency drives a FOXP2-high aggressive state targetable by epigenetic inhibitors

Stem Cell Reports, December 15, 2022, *Dysregulated Polycomb Repressive Complex 2 contributes to chronic obstructive pulmonary disease by rewiring stem cell fate*

16 - Aria Byrd, PhD

Newly Funded Research

Dissecting SCN8A mutant function in epilepsy

Andrew Escayg and colleagues were awarded a March 2023 R01 from the National Institute of Neurological Disorders and Stroke to dissect the function of the voltage-gated sodium channel SCN8A in epilepsy.

This project expands on the Escayg lab's previous work on SCN8A in epilepsy. He proposes to study three different SCN8A variants: R850Q – one of the most severe and recurrent SCN8A

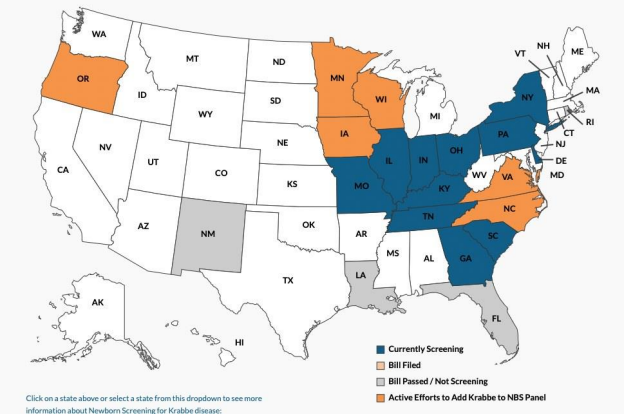
mutations, R1620L – associated with mild epilepsy, intellectual disability and social dysfunction, and N1768D – associated with epileptic encephalopathy. With an eye toward the lack of optimized treatment strategies, researchers will conduct a systematic comparison of selected antiepileptic and candidate drugs for their ability to decrease spontaneous seizures and normalize behavior.

Probing tau pathology in the locus coeruleus

Neuroscience graduate student Anu Korukonda was awarded an F31 NIH predoctoral fellowship from the National Institute on Aging to investigate the behavioral and molecular consequences of tau pathology, and to develop a mouse model in which aberrant forms of tau are exclusively expressed in the locus coeruleus. Korukonda's plans include using translating ribosome affinity purification (TRAP) to isolate the LC transcriptome and probing for adrenergic receptor gene expression in LC-projecting regions of the brain.

17 - Neuroscience graduate student Anu Korukonda

Medical Genetics



18 - Map of the current status of newborn screening for Krabbe disease, from krabbeconnect.org

Hot topic: newborn screening for Krabbe disease

Newborn screening for Krabbe disease, a rare and often fatal leukodystrophy, has been in the news in Georgia lately – thanks to an affected family. A decade ago, one expert called Krabbe "the most controversial newborn screening test in widespread use today" – and it still is.

Starting in 2021, Emory's Department of Human Genetics has been a key participant in a three-year pilot screening program in Georgia for Krabbe disease. How screening occurs here could be a model for other states. But first, some background on why Krabbe is such a heart breaker.

Krabbe disease is caused by a deficiency in the lysosomal enzyme GALC (galactosylceramidase), leading to demyelination and degeneration of the nervous system. Infantile Krabbe usually shows up in the first 6 months of life, with symptoms such as loss of muscle tone, spasticity, irritability, seizures, and loss of head control, hearing, and sight. Most affected children die before age two without intervention. Reviews have concluded that hematopoetic stem cell transplant – the only available treatment – is effective only if performed prior to symptom onset. Even then, transplant carries risks and most long-term survivors "had some degree of functional impairment."

Ten states currently screen for Krabbe, with the others do not. This inconsistency means that based on geography, some cases are identified early in life, while others are identified after the child's decline has become apparent. To speed up diagnosis, affected families and advocacy groups have been pushing for Krabbe disease to be added to the RUSP (Recommended Uniform Screening Panel).

In February 2023, the ACHDNC (Advisory Committee on Heritable Disorders in Newborns and Children) tied when voting on whether Krabbe should be added to the RUSP – and thus Krabbe did not become part of the RUSP. A report prepared for the advisory committee contains current information on Krabbe outcomes.

Two-step screening

Newborn screening for Krabbe disease began in the state of New York in 2006. However, at that point, only a few children who tested positive – based on blood spot analysis for GALC enzyme activity – went on to develop the disease. Extensive neurological testing and follow-up were necessary. In 2015, improved technology became available: testing for blood spot levels of psychosine, the toxic lipid that accumulates in the nervous system in the absence of GALC activity. Georgia's two-step procedure includes a second tier test for psychosine level in samples that show low GALC activity.

Georgia's screening program for Krabbe is going well so far. In the calendar year of 2022, 30 infants screened positive for Krabbe in the first step. In the second step, only one was reported to parents based on elevated psychosine level, according to newborn screening program manager Angela Wittenauer. The family was referred to genetics and Children's Healthcare of Atlanta hematology.

A basic principle of newborn screening policy is that screening should only be undertaken when interventions are available and accessible. With Krabbe, hematopoetic stem cell transplant (HSCT) is not an easy solution, even though some medical centers have accumulated experience with it. When the state of Virginia decided not to perform newborn screening for Krabbe disease, equity concerns about referring infants for HSCT quickly enough were prominent.

Unlike other lysosomal storage disorders, enzyme replacement is not feasible for Krabbe because of the need for the enzyme to pass through the blood-brain barrier. Two companies are developing gene therapy products for Krabbe disease; one of them, Forge Biologics, recently reported that two clinical trials were underway. Forge's gene therapy is given during HSCT, while Passage Bio's is injected into the cisterna magna.

In Other Department News

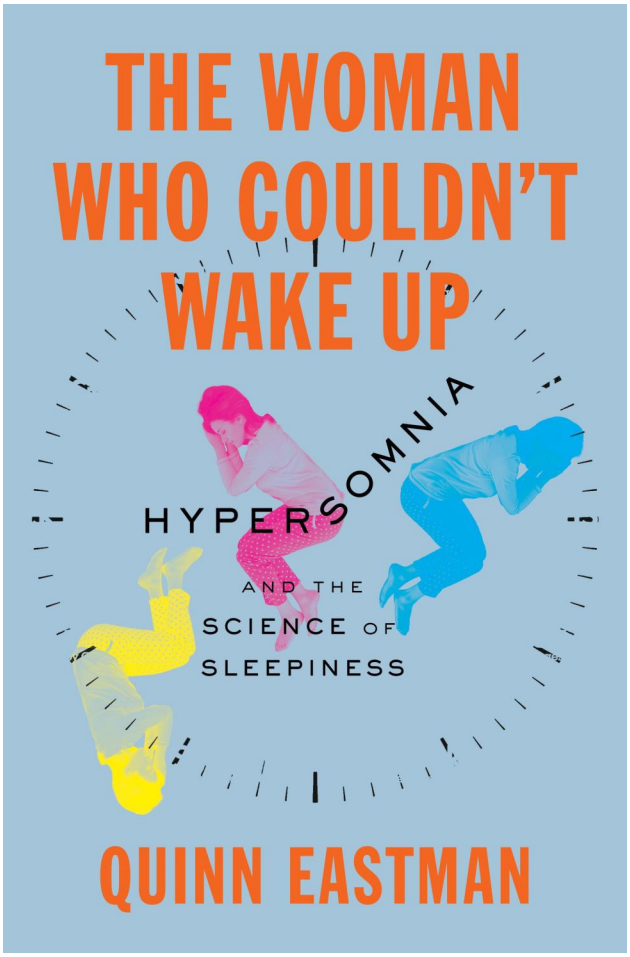
- DOHG Website (link)
- DOHG Events (link)
- More DOHG News (link)

- **SOM News** (link)

Contact Us

DOHG technical editor Quinn Eastman is serving as newsletter editor. To reach him with questions, ideas or submissions for the Human Genetics Dispatch, please email **qeastma@emory.edu**.

Quinn recently completed a book outlining the history of the sleep disorder **idiopathic hypersomnia (IH)**. The Woman Who Couldn't Wake Up was published this summer by Columbia University Press. It covers science and patient advocacy, at Emory and around the world. Genetic studies of IH are underway, and the book contains a proposal to perform exome/genome sequencing analysis on multiplex IH families.



19 - Cover design for The Woman Who Couldn't Wake Up