OCTOBER 2023 | THE PREMED SCENE'S MONTHLY MEDICAL NEWSLETTER

CROSSROADS

THE OFFICIAL NEWSLETTER OF THE PREMED SCENE

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RISING STARS IN MEDICINE: LEAH BURT

Dear medical newsletter readers,

Happy October! We hope you are enjoying the fall weather! Today, we bring you the most updated news in the field of medical research! Ashby Glover is our Rising Stars in Medicine writer, sharing more about Leah Burt's fascinating DCDS learning tool. Then, Mahima Bhat focuses on congenital malformations during fetal growth. Finally, Siri Nikku ends by sharing more about Parkinson's Disease.

Please enjoy reading The Premed Scene's October 2023 Medical Newsletter! Till next month.

Ilana Saidov

Rising Stars in Medicine: Leah Burt

By: Ashby Glover

Leah Burt accepts award from Paul Bergl, fellowship director of Society to Improve Diagnosis in Medicine.

Dr. Leah Burt recently became the first nurse to receive the Emerging Leader ("Rising Star") Award from the Society to Improve Diagnosis in Medicine. Burt was recognized for her creation of the Diagnostic Competency During Simulation-based (DCDS) learning tool, a "first-of-its-kind tool to help improve diagnostic competency within the context of health care simulation." (1) An estimated 40,000 to 80,000 preventable deaths occur per year in U.S. hospitals alone due to missed diagnoses. (2) To combat this, Burt created this educational tool to assess individual competencies in diagnostic reasoning.

"Diagnostic reasoning is an area that has not received enough attention in nursing and medical education. In terms of educating our students, this is critically important because the stakes are so high for our patients." -Leah Burt, PhD '20, MS '10, APRN, ANP-BC. (3)

Using the tool, faculty observe students in a simulated patient encounter, assigning scores to observed behaviors that reflect competencies: "collecting key findings; problem representation; prioritized differential diagnosis; diagnosis justification; decision support; and cognition." (3) For example, for one to be given a high score in differential diagnosis, they would need to include worst-case diagnoses that may be rare, but still essential to have.

The conclusions of her and the research team's pilot experiment demonstrated that the DCDS tool provides nurse practitioner educators with "actionable, competency-specific assessment measures to foster improvement" in diagnostic reasoning. (4)

Burt holds a Ph.D. in Nursing Science from the University of Illinois at Chicago, IL, where she is now a Clinical Assistant Professor and the Director of the AGPC-NP Program. She also maintains an active clinical practice within the University of Illinois Hospital and Health Sciences System's Department of Emergency Medicine.

1. "UIC Nursing faculty member becomes first nurse to receive an award from Society to Improve Diagnosis in Medicine." UIC College of Nursing. October 17, 2023. https://nursing.uic.edu/news-stories/leah-burt-receives-rising-star-award/

2. Lucian Leape, Donald Berwick, and David Bates. "Counting Deaths Due to Medical Errors." Journal of the American Medical Association 288 no. 19 (November 2002): 2405. doi: 10.1001/jama.288.19.2404-jlt1120-2-2

3. "Faculty member creates tool to improve diagnostic skills" UIC College of Nursing. February 27, 2023. https://nursing.uic.edu/news-stories/faculty-member-creates-tool-to-improve-diagnostic-skills/

4. Leah Burt and Andrew Olson. "Development and psychometric testing of the Diagnostic Competency During Simulation-based (DCDS) learning tool" *Journal of Professional Nursing* 45 (2023): 51-59. https://doi.org/10.1016/j.profnurs.2023.01.008

Teratology's Impact on Prenatal Care

By: Mahima Bhat

Teratology is the scientific study of abnormal development, particularly focusing on congenital malformations and the factors contributing to them during embryonic and fetal growth. This field delves into the complexities of how and why abnormalities occur during this crucial development phase, shedding light on the interplay of genetic, environmental, and teratogenic factors.

Inherited genetic changes cause some congenital abnormalities, while others occur due to spontaneous genetic mutations during embryonic development. One key factor is **mutations**, which alter an individual's genetic code. Mutations can occur in critical genes responsible for embryonic development, leading to structural or functional abnormalities in the developing fetus.

Another genetic factor is **inheritance patterns**. Certain congenital abnormalities are inherited in families through specific genetic patterns. For example, autosomal dominant, autosomal recessive, or X-linked inheritance can determine how traits or abnormalities are passed from one generation to the next.

Lastly, **genomic variations** contribute to teratology. Variations in the human genome, including single nucleotide polymorphisms (SNPs) and copy number variations (CNVs), can influence the risk of congenital abnormalities. CNVs and SNPs affect the risk of congenital abnormalities by disrupting the proper dosage of genes involved in embryonic development.

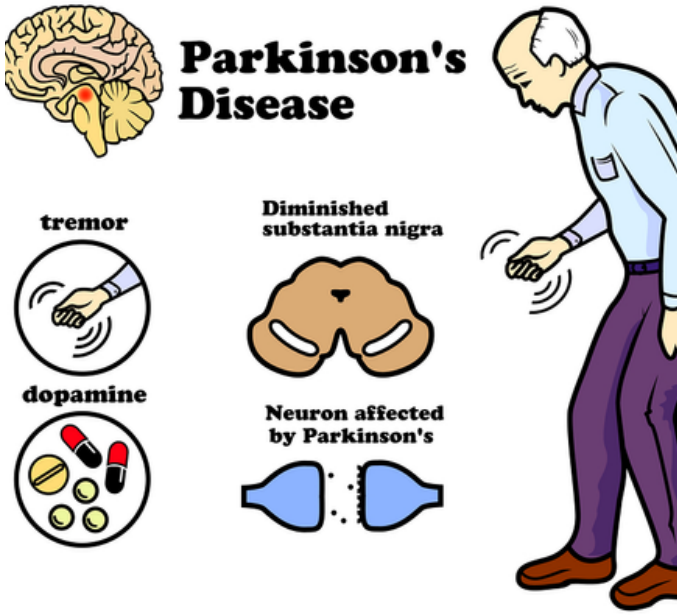
Scientists can now create human induced pluripotent stem cells (iPSCs) to model embryonic development, offering a more ethical and practical way to explore teratogenic effects. Additionally, advancements in computational modeling and bioinformatics have allowed for the prediction of potential teratogenic agents and the assessment of their risks more efficiently.

With the advancement of sophisticated imaging techniques such as 3D and 4D ultrasound, researchers can now visualize embryonic structures in unprecedented detail, allowing for early detection of anomalies. Molecular biology and genetic sequencing have provided valuable insights into the genetic factors of congenital abnormalities, enabling targeted genetic counseling and interventions.

Sources:
https://www.nature.com/articles/2161248a0
https://www.sciencedirect.com/topics/medicine-and-dentistry/teratology

The Causative Genes of Parkinson's Disease

By: Siri Nikku



Parkinson's disease (PD) is categorized as a neurodegenerative disease and differentiated by motor abnormalities such as tremors, slow movements, and rigidity in movements. PD patients often have non-motor symptoms such as orthostatic hypotension, where the blood pressure falls when one is standing up or sitting down, as well as psychiatric symptoms like anxiety or depression. As a result, PD can be defined as a systemic illness and not just entirely the central nervous system being affected.

Along with age, environmental factors like exposure to pesticides and other toxic chemicals can increase the risk of developing PD. There are also genetic backgrounds playing into PD development with two ways to identify it: looking into uncommon Mendelian forms to determine the causative genes and investigating risk factors from genetic statistical analyses of a large group of individuals. Alpha-synuclein gene (SNCA) has been reported as the autosomal dominant form of getting PD. PRKN is another gene that causes PD in the autosomal recessive form for juveniles and is most common in Japan. Two other genes discovered are PTEN-induced kinase 1 (PINK1), which is autosomal recessive, and leucine-rich repeat kinase 2 (LRRK2). While these genes are correlated to the development of PD, there are still many genes that still need to be discovered. Genome-wide association studies (GWAS), an observational study of a genome-wide set of genetic variants in various individuals to see if any variant is connected with a trait, have been used to determine genes. It has shown that in sporadic or irregular intervals, PD has over 200 genes associated with PD development. However, many of these studies have been used mainly for Caucasian populations, not including how ethnicity may play a role in the various forms of the causative genes mentioned. For future work, research will be conducted on how ethnicity, region, and the various genes that cause the development of PD interact with each other to include diverse populations.

Source:
https://www.nature.com/articles/s10038-022-01058-5