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Genetic Competencies for Effective Pediatric Endocrine Nursing Practice☆,☆☆

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Introduction

Our understanding of the molecular basis of health and disease is transforming medicine and nursing. Genetics is highly relevant to pediatric nursing practice as 71% of pediatric admissions have a significant underlying genetic component (McCandless, Brunger, & Cassidy, 2004). In response, nurses need to develop genetic competencies. A number of excellent resources are available to inform nursing practice related to genetics (Behm, 2019). Many nurses may not realize that their skills in history taking and assessment are key components of providing comprehensive genetic healthcare. This article provides an overview of genetics and offers a case example in endocrinology of how genetic competencies can support effective pediatric nursing practice.

Understanding genetics

Genes are like an instruction manual that make you a unique individual. Genes are made up of DNA and are coiled in chromosomes. Genetics refers to how traits (genes) are passed from generation to generation through DNA. Changes in DNA sequence (gene mutations) may occur spontaneously (de novo) or may be passed from parents to offspring. Some genetic changes are gene deletions or insertions, in which genetic material is cut out or extra material is added to a chromosome. Chromosomal disorders are caused by errors during meiosis or mitosis (cell division) and result in autosomal or sex chromosome disorders (Table 1). Epigenetics refers to changes that occur without a change in the DNA sequence (i.e. resulting from lifestyle and environmental factors) (Behm, 2019).

Nurses working in pediatrics need to have a basic understanding of inheritance patterns. Typically, we have two copies of each gene – one inherited from each parent. Autosomal dominant (AD) inheritance is when an affected individual receives an abnormal gene (mutation) from *only one* parent. Examples of AD include achondroplasia (Gene: *FGFR3* locus: 4p16 – i.e. chromosome 4, locus 16) and Marfan syndrome (*FBN1:* 15q21). In autosomal recessive (AR) inheritance, *both* copies of the gene are mutated. An example of AR inheritance is classical congenital adrenal hyperplasia (CAH) (21-hydroxylase deficiency) in which the parents have CAH or are carriers (*CYP21:* 6p21). X-linked inheritance involves a mutation on the X chromosome (a sex chromosome) – and only males (XY) are affected whereas females (XX) are unaffected carriers. An example is X-linked Kallmann syndrome (*ANOS1:* Xp22.31).

History taking

Understanding past medical and family history is vital when trying to gain a comprehensive clinical picture of the child (Fig. 1).

A clinical picture is incomplete without a full, detailed family history. One of the most effective (and least costly) genetic tests involves taking a detailed, 3-generation family history. A family history is essential for identifying individuals who may benefit from genetic services. History taking may evoke strong emotions and thus should be approached in a sensitive and respectful manner. The SCREEN mnemonic is a useful memory aide for guiding family history taking (Table 2) (Trotter & Martin, 2007).

Genetic testing

Clues from the clinical and family history suggesting 'red flag' signs may spur discussion about the utility of genetic testing. Cytogenetic testing can be used to evaluate chromosomal abnormalities while molecular genetic testing is used to identify rare DNA sequence changes (mutations) (Behm, 2019). Best practices include pre-test genetic counselling to support high-quality testing decisions that are informed and aligned with patient values/preferences. In some instances, families may decide not pursue genetic testing and may opt for clinical screening/surveillance programs - particularly if the condition has an adult onset presentation. Notably, genetic results implicate family members so testing decisions may be emotionally charged with psychological implications. Decision-making should be considered a process rather than a singular event, handled thoughtfully and sensitively with a non-judgmental approach. The best interests of the child should remain at forefront of discussion and ethical issues such as consent must be carefully considered (Botkin et al., 2015).

Genetics and endocrinology

Nearly one quarter of findings in genome-wide association studies involve endocrine-related conditions (Goodarzi, 2016). In many

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Table 1
Types and examples of chromosomal disorders in pediatric endocrinology.

Chromosomal error	Description	Clinical Example
Translocation	A 'swap' between two chromosomes	Some cases of growth hormone deficiency and precocious puberty
Deletion	A piece of a chromosome 'breaks off' and is lost	Prader Willi Syndrome: Loss paternally expressed genes (15q11.2 – q13) in 75% of cases
Duplication	A part of the chromosome may be copied more than once	Duplication of Xq26–27 is linked with hypopituitarism

(Adapted from (Behm, 2019)

endocrine consultations, family history taking and genetic testing may inform clinical care. The most common form of CAH is an AR condition of 21-hydroxylase deficiency (*CYP21*: 6p21); the second most common is 11-β-hydroxylase deficiency (*CYP11B1*:8q21) - which also includes hypertension as well as virilizing properties. Some centers advocate for prenatal genetic testing of women and their families to inform pregnancy decisions (Dukhovny & Norton, 2018). For families with CAH, genetic test results may guide decision-making for maternal dexamethasone treatment or pre-implantation genetic testing (Simpson & Rechitsky, 2019). Another potential indication for genetic testing relates to multiple endocrine neoplasia syndromes (Davies, 2018a, 2018b), as well as familial phaeochromocytoma. Genetic testing may help solve diagnostic dilemmas such as differentiating constitutional delay of growth and puberty from abiding hypogonadism (i.e. Kallmann syndrome) (Dwyer, Phan-Hug, Hauschild, Elowe-Gruau, & Pitteloud, 2015). Below we highlight a brief case example demonstrating how sound history taking and genetic testing are central to comprehensive pediatric endocrine nursing practice.

Case study

Tom (pseudonym) presented at 1.8 years following several spontaneous hypoglycemic episodes and severe postnatal growth failure (height velocity < 2 cm/year, −6.96 SDS for height). Birth history revealed he was a fraternal twin born at 35 weeks gestation via caesarean section (length: 53 cm [2.71 SDS], birth weight: 3.3Kg [1.7 SDS] – twin

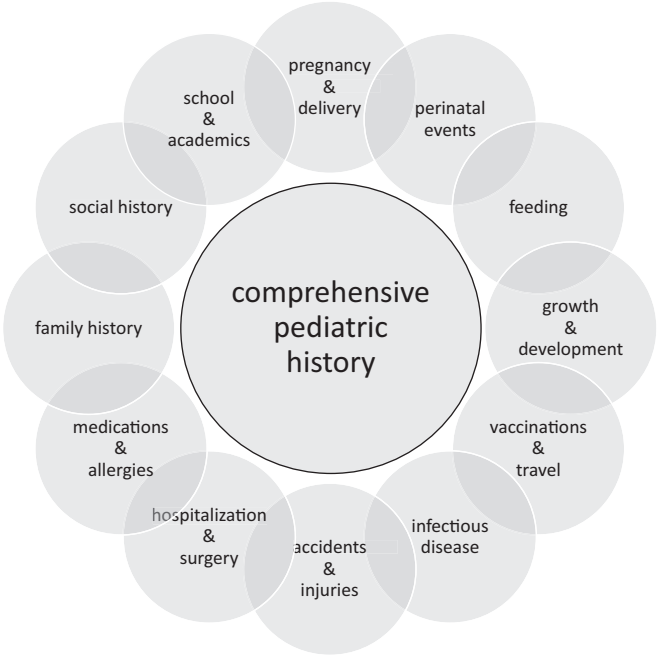

Fig. 1. Components of a comprehensive pediatric history.

Table 2
SCREEN mnemonic for Family History Collection.

SC	**S**ome **C**oncerns	Inquire about any known genetic conditions that run in the family
R	**R**eproduction	Inquire about reproduction problems, i.e. infertility, pregnancy, recurrent miscarriage, birth defects
E	**E**arly disease/death/disability	Inquire about any deaths at a young age, stillbirths, very sick infants (e.g. CAH)
E	**E**thnicity	Inquire about the family ethnic background
N	**N**on-genetic	Inquire about other medical conditions e.g. cardiac, pulmonary, renal

(Adapted from (Trotter & Martin, 2007)

weighed 2.5Kg). Family history indicated unrelated Caucasian parents (British father: 177.8 cm [0.47 SDS], Bahamian mother 175.3 cm [2.21 SDS]). Mutations in *GHR* underlie Laron syndrome and certain ethnic groups and geographic areas appear to have higher concentrations of *GHR* mutations – notably the Bahamas (Baumbach et al., 1997). Thus, the family history elicited a 'red flag'. Following discussion and consent, genetic testing revealed a growth hormone receptor mutation (*GHR:* 5p13-p12) consistent with Laron syndrome. Individuals with Laron syndrome have poor growth, low insulin like growth factor 1 (IGF-1) levels and are unresponsive to growth hormone therapy. Accordingly, Tom was appropriately treated with IGF-1 therapy.

Conclusion

Herein, we summarize some basic genetic principles, demonstrating how genetic competencies (i.e. history taking) is highly relevant to pediatric nursing practice. In this genomic era, nurses must work to integrate genetics into assessment, clinical history taking and nursing practice to provide comprehensive, compassionate care for patients and families.

References

Baumbach, L., Schiavi, A., Bartlett, R., Perera, E., Day, J., Brown, M. R., ... Cleveland, W. (1997). Clinical, biochemical, and molecular investigations of a genetic isolate of growth hormone insensitivity (Laron's syndrome). *Journal of Clinical Endocrinology and Metabolism*, *82*(2), 444–451.

Behm, K. M. (2019). Genetics and family history. In S. Llahana, C. Follin, C. Yedinak, A. Grossman, K. Davies, & M. F. Keil (Eds.), *Advanced practice in endocrinology nursing. Vol. 1*. (pp. 121–161). Switzerland: Springer.

Botkin, J. R., Belmont, J. W., Berg, J. S., Berkman, B. E., Bombard, Y., Holm, I. A., ... McInerney, J. D. (2015). Points to consider: Ethical, legal, and psychosocial implications of genetic testing in children and adolescents. *American Journal of Human Genetics*, *97*(1), 6–21. https://doi.org/10.1016/j.ajhg.2015.05.022.

Davies, K. (2018a). Multiple Endocrine Neoplasia in Children and the Importance of Screening: Part 1. *Journal of Pediatric Nursing*. https://doi.org/10.1016/j.pedn.2018.04.014.

Davies, K. (2018b). Multiple Endocrine Neoplasia in Children and the Importance of Screening: Part 2. *Journal of Pediatric Nursing*, *42*, 129–131. https://doi.org/10.1016/j.pedn.2018.04.015.

Dukhovny, S., & Norton, M. E. (2018). What are the goals of prenatal genetic testing? *Seminars in Perinatology*, *42*(5), 270–274. https://doi.org/10.1053/j.semperi.2018.07.002.

Dwyer, A. A., Phan-Hug, F., Hauschild, M., Elowe-Gruau, E., & Pitteloud, N. (2015). TRANSITION IN ENDOCRINOLOGY: Hypogonadism in adolescence. *European Journal of Endocrinology*, *173*(1), R15–R24. https://doi.org/10.1530/EJE-14-0947.

Goodarzi, M. O. (2016). Genetics of common endocrine disease: The present and the future. *The Journal of Clinical Endocrinology and Metabolism*, *101*(3), 787–794. https://doi.org/10.1210/jc.2015-3640.

McCandless, S. E., Brunger, J. W., & Cassidy, S. C. (2004). The burden of genetic disease on inpatient care in a children's hospital. *American Journal of Human Genetics*, *74*, 121–127.

Simpson, J. L., & Rechitsky, S. (2019). Prenatal genetic testing and treatment for congenital adrenal hyperplasia. *Fertility and Sterility*, *111*(1), 21–23. https://doi.org/10.1016/j.fertnstert.2018.11.041.

Trotter, T. L., & Martin, H. M. (2007). Family history in pediatric primary care. *Pediatrics*, *120*(Suppl. 2), S60–S65. https://doi.org/10.1542/peds.2007-1010D.