

Presented by Eydie Kendall, PT, PhD, PCS

New advances in genetics/genomics and epigenetics/epigenomics have had a bigger transformative effect on healthcare delivery than any other basic science over the past two decades. From the completion of the Human Genome Project to development of pharmacogenetics, precision medicine, and CRISPER-CAS 9, real advances for genetic diseases are here or on the horizon. Since the addition of genetics to the entry-level educational requirements for Doctors of Physical Therapy in 2016, the role and scope of our profession is still being defined. This course provides a review of basic background in genetic concepts, updates on current genetic-based medical interventions, implications for clinical practice with emphasis on pediatrics, and the opportunity to explore PT's role in the implementation of genetics-informed healthcare.

Objectives:

- Describe the recent advances in genetic and epigenetic understanding with respect to movement and disability
- Define some of the major concepts that determine translation from genetic mutation to clinical manifestation
- Apply knowledge of genetics and epigenetics to current clinical practice in pediatrics and other patient populations
- Discuss implications of genetics-based prescriptive healthcare interventions on prognosis and dosage
- Explore the role of physical therapy within the healthcare team for patients with genetic syndromes and conditions

Eydie Kendall is a faculty member with the Doctor of Physical Therapy Program at Plymouth State University. She currently teaches genetic content in two courses, from an organismal adaptation perspective as well as clinically oriented pediatric interventions. She has taught genetics to students in a variety of healthcare, nursing, and rehabilitation programs, for over twenty years. She is a certified clinical specialist in pediatrics and has extensive intervention experience with children with pathologies of genetic etiology.

References:

Ambrosio F, Kleim JA. Regenerative rehabilitation and genomics: frontiers in clinical practice. *Physical therapy*. 2016;96(4):430-432. doi:10.2522/ptj.2016.96.4.430

Cornwall J, Elliott JM, Walton DM, Osmotherly PG. Clinical genomics in physical therapy: where to from here? *Physical therapy*. 2018;98(9):733-736. doi:10.1093/ptj/pzy069

Curtis CL, Goldberg A, Kleim JA, Wolf SL. Translating genomic advances to physical therapist practice: a closer look at the nature and nurture of common diseases. *Physical therapy*. 2016;96(4):570-580. doi:10.2522/ptj.20150112

Duan D, Mendell JR, eds. *Muscle Gene Therapy*. Second ed. Cham, Switzerland: Springer; 2019. doi:10.1007/978-3-030-03095-7

Dunn SL, Olmedo ML. Mechanotransduction: relevance to physical therapist practice-understanding our ability to affect genetic expression through mechanical forces. *Physical therapy*. 2016;96(5):712-721. doi:10.2522/ptj.20150073

Genetic Conditions. National Library of Medicine MedlinePlus. Accessed may 30, 2022. <https://medlineplus.gov/genetics/>

Genomics and precision health. Center for Disease Control. Updated May 18, 2022. Accessed May 21, 2022. <https://cdc.gov/genomics/disease/epigenetics.htm>

Healthcare Provider Genomic Education Resources. NIH National Human Genome Research Institute. Updated May 17, 2022. Accessed May 21, 2022. <https://www.genome.gov/For-Health-Professionals/Providers-Genomics-Education-Resources>

Lake W, Ricks M, Jackman T. Physical therapy interventions and response to treatment in a 13-year-old female with paramyotonia congenita. *Archives of physical medicine and rehabilitation*. 2022;103(3):28. doi:10.1016/j.apmr.2022.01.076

Manolio TA, Bult CJ, Chisholm RL, et al. Genomic medicine year in review: 2021. *The american journal of human genetics*. 2021;108(12):2210-2214. doi:10.1016/j.ajhg.2021.11.006

Pedersen BK. Physical activity and muscle-brain crosstalk. *Nature reviews endocrinology*. 2019;15(7):383-392. doi:10.1038/s41574-019-0174-x

Reilly A, Chehade L, Kothary R. Curing sma: are we there yet? *Gene therapy*. 2022;(20220526). doi:10.1038/s41434-022-00349-y

Reychler G, De Backer M-M, Piraux E, Poncin W, Caty G. Physical therapy treatment of hypermobile ehlers-danlos syndrome: a systematic review. *American journal of medical genetics part a*. 2021;185(10):2986-2994. doi:10.1002/ajmg.a.62393

Rodon LH, Ramocki MB. Chapter 13: Genetics and metabolism in pediatric balance disorders in O'Reilly R, Morlet T, Brodsky JR, Cushing SL, eds. *Manual of Pediatric Balance Disorders*. Second ed. San Diego, CA: Plural Publishing; 2020. INSERT-MISSING-URL.

Sayed N, Allawadhi P, Khurana A, et al. Gene therapy: comprehensive overview and therapeutic applications. *Life sciences*. 2022;294:120375-120375. doi:10.1016/j.lfs.2022.120375

Svendsen CN. Getting the upper hand in ALS. *Gene therapy*. 2022;29(3-4):113-114. doi:10.1038/s41434-022-00314-9

The Human Genome Project. NIH national Human Genome Institute. December 22, 2020. Accessed May 28, 2022. <https://www.genome.gov/human-genome-project>

What's a Genome. NIH National Human Genome Research Institute. Updated October, 11 2019. Accessed May, 30, 2022. <https://genome.gov/About-Genomics/Introduction-to-Genomics>

Woelfel JR, Dudley-Javoroski S, Shields RK. Precision physical therapy: exercise, the epigenome, and the heritability of environmentally modified traits. *Physical therapy*. 2018;98(11):946-952. doi:10.1093/ptj/pzy092