



GROWTH DISORDERS 2025: CURRENT CHALLENGES,
SOLUTIONS AND RECOMMENDATIONS

PODCAST & INFOGRAPHIC

Imprinting Disorders:
A Clinical Guide

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Medical Education Academy

Overview

Ongoing research is revealing the complexities of imprinting disorders, a group of congenital diseases which affect growth, development and neoplasia risk [Mackay D, 2022]. These conditions are caused by genetic or epigenetic changes to genes that are expressed from only one parental allele and can results from changes in coding sequences, copy number changes, uniparental disomy or imprinting defects [Mackay D, 2022]. Specifically, imprinting disorders are characterized by molecular alterations affecting differentially methylated regions (DMR) [Eggermann T, 2022]. Individuals with a particular imprinting disorder, in general, have a single specific affected DMR, but individuals can have multi-locus imprinting disturbances (MLID) [Eggermann T, 2022]. Individual imprinting disorders, such as Prader-Willi syndrome, Angelman syndrome, Silver-Russell syndrome and Beckwith-Wiedemann syndrome, have similar clinical features but are heterogeneous, with key clinical manifestations often non-specific, making diagnosis difficult [Eggermann T, 2023]. Research is ongoing into the regulation within DMRs and their functional consequences as well as the cross-talk between imprinted genes and functional pathways.

Learning Objectives

At the end of the podcast, learners should be able to:

1. Understand, differentiate and describe imprinting disorders
2. Be aware of the underlying genetic mechanisms resulting in these disorders
3. Summarize the diagnosis and management of imprinting disorders

Target Audience

Paediatricians working in growth disorder management and those in general practice; specialist growth disorder nurses.

Language

English with voice-over into Spanish and Korean.

Faculty



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Programme



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