

VIDEO INTERVIEW - New genetic causes of growth disorders

OVERVIEW

Growth failure is the most common reason for referring children to a secondary or tertiary medical center, and to pediatric endocrinologists especially. Genetic disorders account for a substantial part of the underlying causes. Scientific progress has much improved the knowledge on the mechanisms regulating human growth and on potential genetic defects affecting stature in childhood. Several new genetic techniques have been developed and delivered worldwide, permitting a better investigation of many growth disorders which were defined as idiopathic until a few years ago. Gene defects have been described affecting both GH-IGF1 axis and bone growth plate, as well as other signaling pathways. Based on this, it is crucial for health-care professionals to remain updated on the molecular disorders causing short stature and to be confident with the most modern genetic techniques for investigating them appropriately. We propose an interview with one of the most famous international experts on genetic disorder causing poor growth in children. Prof. Horacio Domené was first in describing ALS deficiency related short stature. He will share with participants the story of his career, from the beginnings until present days, providing precious advices on how to approach to gene analysis for short stature patients.

LEARNING OBJECTIVES

1. How to start a successful career in pediatric endocrinology
2. How first evidence of ALS mutation related short stature came up to the light
3. When genetic investigations are needed in children with growth failure
4. Genes currently known to be involved in growth disorders
5. Limitations of genetic tests available nowadays and development of new technologies to address the current issues.

TARGET AUDIENCE

This activity is intended for all healthcare providers involved in the care of diseases affecting growth in childhood, in particular specialist care providers, including pediatric endocrinologists, scientists, pediatricians involved in chronic disease care and pediatric nurses.



FACULTY

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