



CHILD NEUROLOGY: KEY UNDERSTANDINGS AND INTERVENTIONS FOR CHILDREN WITH DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHIES

HOW TO IDENTIFY AND MANAGE METABOLIC CAUSES OF DEE

Vincenzo Leuzzi

Unit of Child Neurology and Psychiatry

Department of Human Neuroscience

Sapienza – Università di Roma

vincenzo.leuzzi@uniroma1.it



SAPIENZA
UNIVERSITÀ DI ROMA

DISCLOSURES

No potential conflict of interest to report

LEARNING OBJECTIVES

- To define the concept of metabolic and developmental epileptic encephalopathies (MDEE)
- To acquire an etiological classification of MDEE
- To identify the key clinical features suggesting a MDEE
- To learn and master biomarkers of MDEE focusing on those requiring a specific and disease modifying treatment
- To learn the correct diagnostic work-up for a rapid identification of MDEE

KEY MESSAGE

- A relatively restricted number of Developmental Epileptic Encephalopathies (DEE) are due to genetic alterations affecting proteins involved in metabolic pathways.
- Although minority in comparison to other causes of DEE, some of these conditions offer the advantage, if early detected, of a specific treatment which can modify course and outcome of the disease.
- Clinicians should pay particular attention to these metabolic DEE, which can be identified on the base of clinical presentation and specific metabolic biomarkers.
- This course provides the basic knowledges for a correct approach to child with DEE in order to address the diagnosis of metabolic DEE.

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