



ChildCA Project, WP4: Training Course in Genetics

Speaker
**Prof. Enza Maria
Valente**

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www.childca.eu



childca@unipv.it



Prof. Enza Maria Valente

- Full professor of medical genetics at the Department of Molecular Medicine of the University of Pavia and head of the Specialized Section of Medical Genetics at the Mondino Foundation IRCCS;
- prof. Valente's research focused on the identification of new disease genes and the mutational analysis of large case series using conventional techniques and next-generation sequencing, genotype-phenotype correlations and characterization of the molecular and functional mechanisms underlying numerous hereditary diseases;
- the author of over 200 scientific publications in peer-reviewed international journals (overall IF: 1396.5 H-index Scopus: 56, over 16,000 citations).

Genetics: Course Structure

1. First Level (Basic Course)

- > Delivery of pre-recorded classes
- > Live session to discuss doubts on the first set of topics

2. Second Level (Advanced Course)

- > Delivery of pre-recorded classes
- > Live session to discuss doubts on the first set of topics



Genetics: Course Structure

Basic lectures

1. Basic concepts
 2. Mendelian genetics
 3. Penetrance and expression
 4. Chromosomal number anomalies
 5. Chromosomal structural anomalies
 6. Mitochondrial genetics
 7. Dynamic mutations
 8. Multifactorial disorders
 9. De novo variants and mosaicism
- + **Live Session**

Genetics: Course Structure

Advanced lectures

1. Indications to genetic testing
 2. Choice of genetic testing
 3. Interpretation of genetic testing and testing asymptomatic subjects
 4. Prenatal and preimplantation diagnosis
 5. Introduction to Next generation sequencing techniques
 6. NGS bioinformatic analysis
 7. Mutation nomenclature
 8. The non-coding genome
 9. Basic notions of genetic therapy
- + **Live Session.**