//CONVEGNI

Parkinson Frontiers: focus on GBA1

Seminar - Thursday 19th March 2026

Sala Gerola, Castello del Buonconsiglio,
via Bernardo Clesio, 5
Trento - Italy

Course - Friday 20th - 21st March 2026

MuSe - Museo delle Scienze
Corso del Lavoro e della Scienza, 3
Trento - Italy

Scientific committee

Maria Chiara Malaguti
Department of Neurology Asuit, Santa Chiara Hospital, Trento, Italy.
Coordinator of the Parkinson's Clinical Network, Asuit Trento, Italy

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Alessio Di Fonzo

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PROVINCIA AUTONOMA DI TRENTO

Introduction

The third edition of PD Frontiers focuses on Parkinson's disease associated with GBA1 gene mutations-one of the most dynamic and promising areas in modern neurology.

The decision to dedicate the course to GBA1-related Parkinson's disease reflects thestrategic importance of this subgroup. Variants of the GBA1 gene represent the most common genetic risk factor for Parkinson's disease, found in a significant proportion of patients (5-15%, reaching up to 25% in specific cohorts).

Beyond its epidemiological relevance, this form stands out for its unique clinical and prognostic characteristics, which call for a dedicated diagnostic and therapeutic approach.

Recent evidence on pathogenic mechanisms-including lysosomal dysfunction, α-synuclein accumulation, oxidative stress, and mitochondrial deficits-has opened new therapeutic avenues that are now the focus of numerous advanced-stage clinical trials.

The scientific value of the course is ensured by the participation of leading national and international experts in genetics, neurobiology, and innovative therapies for GBA1-related Parkinson's disease.

Featuring both national and international lecturers, the seminar and the course offer participants a unique opportunity for up-to-date learning, critical discussion, and sharing of the latest scientific evidence.

This highly stimulating environment promotes dialogue among experts from complementary disciplines, helping to build a truly global scientific community dedicated to advancing knowledge and improving care for GBA1-related Parkinson's disease.

Seminar

Sala Gerola, Castello del Buonconsiglio,
via Bernardo Clesio, 5 - Trento

19th March 2026

13.00 Registration

13.45 Opening Authorities

14.15 Introduction to the seminar
Scientific Directors: Maria Chiara Malaguti, Alessio Di Fonzo

Chairmen: *M. C. Malaguti, A. Di Fonzo*

14.30 Genetic and molecular mechanism linking GBA1 and PD across different populations
A. Di Fonzo

15.05 Biochemical assessment and lipid storage in Gaucher disease
J. Aerts

15.40 Impact of PD-GBA1 in Italy
E. M. Valente

16.15 Coffee break

Chairmen: *M. Marano, C. Zizzo*

16.45 Parkinsonism in Gaucher Disease patients and GBA1 carriers
A. Zimran

17.20 GBA 1 and LRRK2 mutations in fly models
M. Horowitz

17.55 Targeted therapies for GBA1 PD
R. Cilia

18.30 Closing remarks

18.45 Closure

Course

MuSe - Museo delle Scienze,
C.so del Lavoro e della Scienza, 3 - Trento

20th March 2026

FOCUS ON MOLECULAR-CLINICAL CORRELATION

08.30 **Registration**

09.00 **Introduction**
Course directors: Maria Chiara Malaguti, Alessio Di Fonzo

09.15 **Lysosome dysfunction in neurological disorders**
G. Duro

09.45 **Biochimical Focus**
C. Zizzo

10.15 **Genetics Focus**
E. Monfrini

10.45 **Coffee break**

11.15 **QSP model in lysosomal storage disease**
F. Reali

11.45 **Clinical Focus**
M. Marano

12.15 **Non neurological phenotype of GBA1 patients**
I. Motta

12.45 **Lunch**

14.00 **Clinical series**
A. Di Fonzo, D. Ottaviani

15.30 **Working groups (parallel sessions):**

Genetics:

- How to classify GBA1 mutations in PD
  A. Di Fonzo
- Type of Technology to detect GBA1 variants
  A. E. Dardis

Biochemical:

- How to measure GCase activity in patients
  C. Zizzo
- Challenges in interpretation of biochemical results in patients
  N. Vitturi

Clinical:

- Standardized neurological phenotyping of GBA1 PD (device e cognitivo)
  A. Magliozzi
- Brain Imaging in GBA1 patient
  F. Cavallieri

16.30 **Video challenge**

17.00 **Poster session**

18.20 **Closing remarks**
A. Di Fonzo

18.30 **Closure**

21st March 2026

FOCUS ON THERAPIES IN GBA1 PD

09.00 **Symptomatic therapy**
G. Di Lazzaro

09:30 **Infusion therapies**
C. A. Artusi

10.00 **Deep brain stimulation**
M. Avenali

10.30 **Coffee Break**

11.00 **Treatment of dysautonomia in parkinsonian syndromes**
A. Fanciulli

11.30 **Non pharmacological treatment**
M. Putzolu

12.00 **Future perspectives**
A. Di Fonzo

12.30 **Closing remarks**

12.45 **Closure**

Faculty

Aerts Johannes

Professor Hans (JMFG) Aert chaired the Dep. Medical Biochemistry at Leiden University and at the AMC-Amsterdam (2000-2023). As Emeritus, he still supervises 5 PhD students and a postdoctoral fellow. His research focuses on glycosphingolipids in health and disease with special attention for inherited lysosomal storage disorders like Gaucher disease (GBA-PD) as well as neurodegeneration. He elucidated the life cycle of GCase and was involved in the first applications of enzyme replacement therapy and substrate reduction therapy of Gaucher disease as well as the discovery of widely used biomarkers. He identified novel substrates for GBA1 as well as GBA2, and developed small compounds to visualize and inhibit the cellular β-glucosidases (GBA1 and GBA2), with potential therapeutic and diagnostic applications. He has been promotor of 56 completed PhD theses and published >450 peer-reviewed papers (H-index: 107, Google Scholar).

Artusi Carlo Alberto

I am an Associate Professor of Neurology at the Department of Neuroscience, Biomedicine and Movement at the University of Verona, Italy, where I work as a senior neurologist and scientific researcher in the field of neurodegenerative diseases, with a particular focus on Parkinson's disease and other movement disorders, advanced therapies for Parkinson's disease, postural abnormalities in parkinsonism, mobile health technology (mHealth), Lewy body dementia and other forms of cognitive impairment associated with Parkinson's and parkinsonism, and the use of botulinum toxin in movement disorders. I was a member of the MDS Task Force on Postural Abnormalities in Parkinsonism and I am a member of the Steering Committee of the MDS "Gait and Posture" study group. I was also a member of the Technical Scientific Committee of the LIMPE Foundation for Parkinson's Onlus and coordinator of the youth group of the Italian Society for Parkinson's and Movement Disorders/LIMPE-DISMOV.

Avenali Micol

Senior Assistant Professor of Neurology, Dep. of Brain and Behavioural Science, University of Pavia, Pavia - Italy; Parkinson and Movement Disorders Unit, Area of Neurodegenerative and Adult Rare Diseases, IRCCS Mondino Foundation, Pavia, Italy. Dr. Micol Avenali, MD, PhD, is a Clinical Neurologist and Senior Assistant Professor of Neurology at the IRCCS Mondino Foundation and University of Pavia. She specializes in neurodegenerative disorders, with a focus on genetic movement disorders, including Parkinson's disease. Dr. Avenali coordinates the Deep Phenotyping Research Unit at Mondino and leads studies on the role of GBA mutations, integrating clinical, biochemical, and imaging biomarkers to define genotype-phenotype correlations. Her work also emphasizes advanced therapies, including deep brain stimulation (DBS) and other interventional approaches for movement disorders. She holds national and international collaborations with leading centers in Movement Disorders and Neurogenetics. Her research outputs include high-impact publications and competitive grants.

Cavallieri Francesco

MD, PhD, is the Head of the Movement Disorders Center and Consultant Neurologist at the Neurology Unit of the Arcispedale Santa Maria Nuova – AUSL IRCCS of Reggio Emilia, Italy. He obtained his MD and specialization in Neurology with honours at the University of Modena and Reggio Emilia, where he also earned a PhD in Clinical and Experimental Medicine. His clinical and research activity focuses on Parkinson's disease, with specific expertise in advanced therapies and multimodal clinical phenotyping. His research is particularly dedicated to genetic forms of Parkinson's disease, with a main focus on GBA-associated Parkinson's disease, including genotype - phenotype correlations and disease modifiers. A key area of interest is the instrumental and neuroimaging assessment of motor and non-motor features in GBA1-PD, aimed at improving stratification and personalized therapeutic approaches.

Cilia Roberto

Has been Head Neurologist at S.C. Neurologia 1 – Parkinson's and Movement Disorders at the I.R.C.C.S. Carlo Besta Neurological Institute since November 2019. Since August 2023, he has been Head of the Parkinson's and Continuity of Care Unit. From a scientific point of view, he is involved in clinical research, mainly focused on drug therapy and the pathophysiology of Parkinson's disease. He has published over 150 scientific articles with an h-index of 45. He is the Scientific Director of several studies, including a double-blind randomized clinical trial sponsored by the Ministry of Health on the effects of Ambroxol as a disease-modifying therapy in patients with Parkinson's disease and GBA1 gene mutations. He has received international awards such as the Junior Award from the Movement Disorder Society for Clinical Research in June 2010 (Buenos Aires) and the "2016 Excellence in Neurology" award (Toulouse, France). He is Associate Editor of the journal "Parkinsonism and related Disorders," published by Elsevier.

Dardis Andrea Elena

Position: Head of the laboratory of the Regional coordinator Centre for Rare Diseases Institution: University Hospital of Udine. Dr. Andrea Dardis obtained a MS degree in Biochemistry and then a PhD in Molecular Biology at the University of Buenos Aires, Argentina. She continued her training at the Metabolic Unit, University of California, San Francisco, USA as a post-doctoral fellow. During her training she was awarded the International Fellowship of the Lawson Wilkins Pediatric Endocrine Society. She got a Specialist Degree in Medical Genetics at the University of Genoa. In 2003 she joined the Metabolic Diseases Unit, Pediatric Hospital Trieste, Italy as a Research Scientist. In 2009 she became Head of the Laboratory at the Regional Coordinator Centre for Rare Diseases in Udine, Italy,. She is responsible for the "Biobank of ALS, Neuromuscular and Lysosomal Diseases" of the University Hospital of Udine and member of the Board of Trustees of the International Niemann Pick Disease Registry, Vice-Chair of the International Working group of Gaucher Disease (IWGGD) and Council member of the Society for the Study of Inborn Errors of Metabolism (SSIEM). Her activities are mainly focused on the biochemical and molecular diagnosis of lysosomal storage diseases, the functional characterization of defective lysosomal enzymes and the pre-clinical studies of novel therapeutic for lysosomal storage disorders.

Di Fonzo Alessio

IRCCS Foundation Cà Granda Ospedale Maggiore Policlinico, Dino Ferrari Center, Neuroscience Section, Department of Pathophysiology and Transplantation, University of Milan, Italy. Alessio Di Fonzo is a board-certified neurologist trained as a movement disorders specialist. His activity comprises the in-patient clinic, and the direction as Principal Investigator of the out-patient clinic for Movement Disorders and Rare Neurological Diseases. He is the Director of the Movement Disorder Group, Fresco Institute of Milan. In the last 5 years, he directed his group's research focus on genetics of Parkinson's Disease, investigating both the molecular and clinical aspects. His team is composed of excellent young researchers (neurologists and biologists) specialized in the field of neurogenetics and directed towards a promising academic career. The principal research subjects are: 1) Genetics of Movement Disorders; 2) iPSCs-based cellular models of PD; 3) Gaucher Disease and GBA1-PD.

Di Lazzaro Giulia

Graduated in Medicine and Surgery (2014) at Catholic University of Rome, tutored by prof. Anna Rita Bentivoglio. She then completed the Neurology residency (2019) and the Neuroscience PhD (2023) at Tor Vergata University of Rome, with particular interest in movement disorders, tutored by prof. Antonio Pisani and prof Paolo Calabresi. She spent 18 months at Queen Square institute of neurology as a research fellow, tutored by prof. Kailash Bhatia between 2019 and 2020 as winner of the EAN research fellowship. She now works as consultant neurologist at Fondazione Policlinico Universitario Agostino Gemelli IRCCS and is a contract professor at Università Cattolica del Sacro Cuore in Rome. She also tutors medicine students and Neurology residents. She has published in peer reviewed international journals (h index 22), participated to national and international congresses with posters and oral communications, and collaborates in multicenter national and international projects. She speaks fluently Italian, English, French and Spanish.

Duro Giovanni

Research Director - IRIB-CNR Palermo. Head: Center for Research and Diagnosis of Lysosomal Storage Diseases, IRIB CNR. Adjunct Professor, Lecturer in Genetics of Lysosomal Storage Diseases - School of Medicine – UniPA. Our center studies and diagnoses the following disorders: Fabry; Pompe; Gaucher; Nieman-Pick; MPS1.

Fanciulli Alessandra

Is Associate Professor of Neurology and Director of the Dysautonomia Center at the Department of Neurology, Medical University of Innsbruck. Her research focuses on autonomic failure and other non-motor symptoms in parkinsonian and related neurodegenerative disorders. She coordinates an FWF-funded research consortium on medical decision-making in multiple system atrophy (MSA) and leads studies on pain in people living with MSA. Prof. Fanciulli serves as President of the European Federation of Autonomic Societies and of the Bischof Karl Golser Foundation for Research in Atypical Parkinsonian Syndromes. She chairs the MSA Study Group of the International Parkinson and Movement Disorder Society and the Atypical Parkinsonian Syndromes Disease Group of the European Reference Network for Rare Neurological Disorders. For the European Academy of Neurology, she coordinates the forthcoming guidelines on the symptomatic treatment of atypical parkinsonian syndromes. In 2020, she was awarded the Bilateral Scientific Cooperation Award by the Italian Ministry of Foreign Affairs. Most importantly, she is the proud mother of three wonderful boys.

Horowitz Mia

Professor, Shmunis School of Biomedicine and Cancer Research, Life Sciences, Tel Aviv University. I have been studying the molecular biology of Gaucher disease (GD) for over 30 years. My lab cloned and sequenced the GBA1 gene, defective in GD and its closely related pseudogene. We showed that mutant GCase is retained in the ER, triggering the Unfolded Protein Response (UPR). Using Drosophila

melanogaster as an animal model, our research showed that expressing human mutant GBA1 variants in flies induces parkinsonian signs. In recent years, we have also explored the interplay between mutant GBA1 and mutant LRRK2 alleles and found that mutant LRRK2 reduces the steady-state levels of mutant GBA1, thereby alleviating parkinsonian symptoms in the fly model.

Magliozzi Alessandro

Is a board-certified neurologist and attending physician at the Neurology Unit, ASST Spedali Civili di Brescia, Italy. His clinical activity focuses on Parkinson's disease and movement disorders, including advanced and device-aided therapies. His research interests focus on the genetic basis of Parkinson's disease, with particular emphasis on GBA-associated forms. He is currently a PhD candidate in Neuroscience, with a translational focus on biomarker-driven precision medicine in Parkinson's disease.

Malaguti Maria Chiara

MD, is a board-certified Neurologist currently serving as Senior Consultant at the Neurology Unit, Santa Chiara Hospital, Trento (Asuit, Italy). She holds a senior clinical leadership role in the clinical management of Parkinson's disease and coordinates the Provincial Parkinson Clinical Network. Her main areas of expertise include movement disorders and advanced therapies for Parkinson's disease (deep brain stimulation and infusion therapies). She is actively involved in national and international research on neurodegenerative diseases, genetics, and artificial intelligence in clinical neurology. Dr. Malaguti holds leadership roles within scientific societies, including the European Academy of Neurology, as co-chair of the EAN Task Force on Artificial Intelligence in Clinical Neurology. Her educational interests focus on multidisciplinary care, digital health, and innovation in continuing medical education.

Marano Massimo

Neurologist at the Campus Bio-Medico University Hospital Foundation in Rome, where he coordinates the Parkinson's and Movement Disorders Centre.
His main clinical activity is in the diagnosis and treatment of early-stage or complicated Parkinson's disease, where he manages the use of advanced infusion therapies and deep brain stimulation. The Campus research group focuses on clinical and neurophysiological research, with particular attention to the genetic and phenotypic characterisation of GBA Parkinson's disease and movement disorders, thanks to collaboration with the Policlinico di Milano, Asuit in Trento and the CNR IRIB in Palermo.

Monfrini Edoardo

Is a consultant neurologist at the Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico in Milan, Italy. He graduated with honors in Medicine and Surgery (2015), completed his Neurology residency with honors (2020), and earned a PhD in Clinical Research (2022). In 2025, he completed a Master's degree in Genomic Data Science at the University of Pavia, Italy. His clinical and research interests focus on movement disorders and rare neurogenetic diseases. In 2021, he was a Fulbright visiting researcher at NYU Langone (USA), working on innovative genetic diagnostic programs. Has authored over 80 peer-reviewed publications, contributing to the discovery of novel genetic causes of dystonia, ataxia, and parkinsonism. He has been an invited speaker at national and international meetings and is an active member of collaborative research networks such as PARKNET and the Italian Virtual Institute Network for Neuroscience. His research integrates clinical, genetic, and bioinformatic approaches to unravel the molecular basis of neurodegenerative diseases and to improve early and personalized diagnosis.

Motta Irene

I am an Associate Professor of Internal Medicine at the University of Milan, Milan (Italy) and the medical provider for over 500 patients with rare diseases at Fondazione IRCCS Ca' Granda Policlinico in Milan. I'm the director of the research team of the Hemoglobinopathies and Congenital Disorders of Metabolism and Immunity Center in the Unit of Medicine and Metabolic Disease at the aforementioned Institute. During my residency in Internal Medicine, I spent a year (2014) as a Research Fellow in Dr. Stefano Rivella's lab at the Weill Cornell Medical College of Cornell University in New York City (USA). My research endeavor has then led me to advance studies in collaboration with laboratories focused on the cure for rare diseases, hemoglobinopathies, and lysosomal storage disorders. As an Internist, my activities and research interests include clinical management, emerging complications, and new treatment options for patients with hemoglobinopathies and lysosomal storage disorders.

Ottaviani Donatella

MD, PhD, is a board-certified neurologist and currently serves as Senior Consultant at the Neurology Unit of Santa Maria del Carmine Hospital in Rovereto (Asuit, Italy). She leads the Movement Disorders Clinic and the Botulinum Toxin Clinic, operating within the Italian Botulinum Toxin Network (RITB). Her clinical activity focuses primarily on Parkinson's disease and dystonia. She specializes in neurodegenerative disorders. During her residency, she spent nine months at the Institute of Neurology and Neurosurgery, University of London, and at Middlesex Hospital, where she worked as an Honorary Clinical Assistant to Prof. A. J. Lees in 2003. Dr. Ottaviani obtained her PhD in Clinical and Experimental Neurosciences from Sapienza University of Rome in 2010.

Putzolu Martina

Is a Tenure-Track Researcher at the Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics and Maternal and Child Sciences of the University of Genoa, Italy. She holds a PhD in Neuroscience and has a background in Physiotherapy. She has held international experiences at KU Leuven, collaborating with leading experts in movement disorders. Her research focuses on the neurophysiology of movement and neurorehabilitation in Parkinson's disease, with a particular focus on gait disorders and freezing of gait. She investigates innovative non-pharmacological rehabilitation approaches, including motor imagery, action observation, and neuromodulation techniques.

Reali Federico

PhD, is Group Leader of the Quantitative Systems Pharmacology (QSP) group at Fondazione The Microsoft Research – University of Trento Centre for Computational and Systems Biology (COSBI) in Rovereto, Italy. His expertise is in systems biology and QSP modeling applied to drug development across multiple therapeutic areas, including neurodegenerative disorders and lysosomal storage diseases, with specific applications to Parkinson's disease and GBA-related forms. He leads and coordinates multidisciplinary research teams and collaborates with pharmaceutical companies and non-profit organizations on translational QSP projects. He is also a Teaching Fellow at the University of Trento, where he teaches probability and statistics and supervises undergraduate and graduate students. Federico Reali serves as a member of the Scientific Committee of the Italian National Life Sciences Cluster ALISEI. His current interests focus on using quantitative models, digital twins, and data-driven approaches to support drug development, clinical decision-making, therapeutic stratification, and the translation of complex biological data into clinically meaningful insights.

Valente Enza Maria

MD, PhD, is a neurologist with a PhD in Neurogenetics. She is professor of Medical Genetics at University of Pavia and coordinates the Area of Neurodegenerative and Adult Rare Diseases at the IRCCS Mondino Foundation, in Pavia, Italy. One of her main interests is Parkinson Disease (PD), with a focus on translational research. She contributed knowledge on the mutational spectrum, phenotypes and molecular functions of several PD-genes, with focus on PINK1, GBA1 and SNCA. She coordinates the National Virtual Institute of Parkinson Disease (IRCCS Network for Neuroscience and Neurorehabilitation), and is co-lead of the Data Generation Working Group, within GP2 Monogenic Network (www.GP2.org).

Vitturi Nicola

MD Ph.D. Positions I have held various positions at the University Hospital of Padova, Italy, contributing to the field of metabolic diseases and internal medicine. From 2012, I have worked at the Division of Metabolic Diseases of the University Hospital of Padova as Dirigente Medico until today.
Additionally, from 2023 I held a professional assignment for High Specialization in Rare Metabolic Diseases of Adults within the same division. From 2018, I am the Coordinator of the Rare Metabolic Diseases of Adults Service at the University Hospital of Padova, overseeing long-term clinical care, multidisciplinary team coordination, and advanced diagnostic protocols. Institutional responsibilities 2023-today: member of the Scientific Committee of the patient advocacy association 'Italian Anderson-Fabry Association' (AIAF) 2021-today; member of the Adult Metabolic Working Group of the Italian Society for Inherited Metabolic Diseases and Neonatal Screening (SIMMESN) and of the Adult Metabolic Physician Group of the Society for the Study of Inborn Errors of Metabolism (SSIEM).

Zimran Ari

MD, is the founder and former director of the Gaucher Unit at Shaare Zedek Medical Center, Jerusalem, where he currently serves as a senior physician. The Gaucher Unit is the world's largest referral clinic for this genetic disease, following nearly 1,000 patients, half of whom receive specific treatment withintravenous enzyme replacement or oral substrate reduction therapy. He has authored over 360 scientific papers and edited three books. Prof Zimran was first to report the relationship between GD and Parkinson (GBA1-related PD; Sidransky syndrome), and has recently founded AGYANY Pharma focusing on high-dose Ambroxol as chaperone therapy for both GD and Sidransky syndrome.

Zizzo Carmela

Is a Senior Researcher at the National Research Council (CNR), specializing in genetics and lysosomal storage diseases. She is based at the Institute for Research and Biomedical Innovation (IRIB-CNR) in Palermo, Italy, where she leads a research group focused on Gaucher disease and Acid Sphingomyelinase Deficiency (ASMD).
Dr. Zizzo's work investigates genetic and biochemical alterations in glucocerebrosidase, the enzyme responsible for Gaucher disease.
Her research aims to advance our understanding of lysosomal function and the molecular mechanisms underlying these rare genetic disorders.

Informazioni

Il seminario si terrà in lingua inglese.

Destinatari

Seminario: neurologi, ricercatori (biologi/biotecnologi), genetisti, ematologi, internisti.
Corso residenziale: neurologi, ricercatori (biologi/biotecnologi), genetisti.

Iscrizione

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Info ECM

Seminario: 4 crediti ECM
Corso: 11 crediti ECM

Obiettivo dossier formativo cogeaps: tecnico-professionale.
Area tematica: Clinico-Assistenziale - Aggiornamenti nelle discipline specialistiche di competenza.
Obiettivo nazionale: 18. Contenuti tecnico-professionali (conoscenze e competenze) specifici di ciascuna professione, di ciascuna specializzazione e di ciascuna attività ultraspecialistica, ivi incluse le malattie rare e la medicina di genere.

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1. Registrazione delle presenze tamite foglio firma ai fini ECM.
2. Per i dipendenti Asuit l'evento si qualifica come formazione interna. La partecipazione è in orario di servizio in TIMBRATURA causalizzata "formazione interna" con codice 90000.

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Il rilascio dell'attestato è subordinato alla partecipazione al 90% delle ore previste dal programma e al superamento della valutazione di apprendimento.

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Trento
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