



Institute of Neurological Science

The ISN provides highly specialized diagnostic services of molecular genetics and neuroimaging for a wide range of nervous system hereditary diseases. It represents the only CNR Institute authorized by the Italian National Health Service.

#### GENETIC DIAGNOSTIC SERVICES

Aceruloplasminemia  
Cerebral autosomal dominant  
arteriopathy with subcortical infarcts and  
leukoencephalopathy  
Ataxias  
Atrophies  
Neuronal ceroid lipofuscinosis  
Huntington disease  
Familial fronto-temporal dementia  
Dystrophies  
Epilepsy  
X-linked lissencephaly type 1-SCLH  
Alzheimer disease  
Charcot-Marie-Tooth disease  
Fabry disease  
Parkinson disease

Tay Sachs disease  
Mitochondrial diseases  
Neuroacanthocytosis  
Pantothenate kinase-associated  
neurodegeneration  
Neurodegeneration with brain iron  
accumulation, type 2  
Neurofibromatosis  
Familial amyloid polyneuropathy  
Hereditary neuropathy with liability to  
pressure palsies  
Hereditary spastic paraparesis  
Amyotrophic lateral sclerosis  
Rett syndrome  
Startle disease  
Familial thrombophilia

#### RADIOLOGY SERVICES

Standard and advanced magnetic resonance imaging

**ISN**  
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National Research Council of Italy

[www.isn.cnr.it](http://www.isn.cnr.it)  
[www.cnr.it](http://www.cnr.it)

#### CONTACTS

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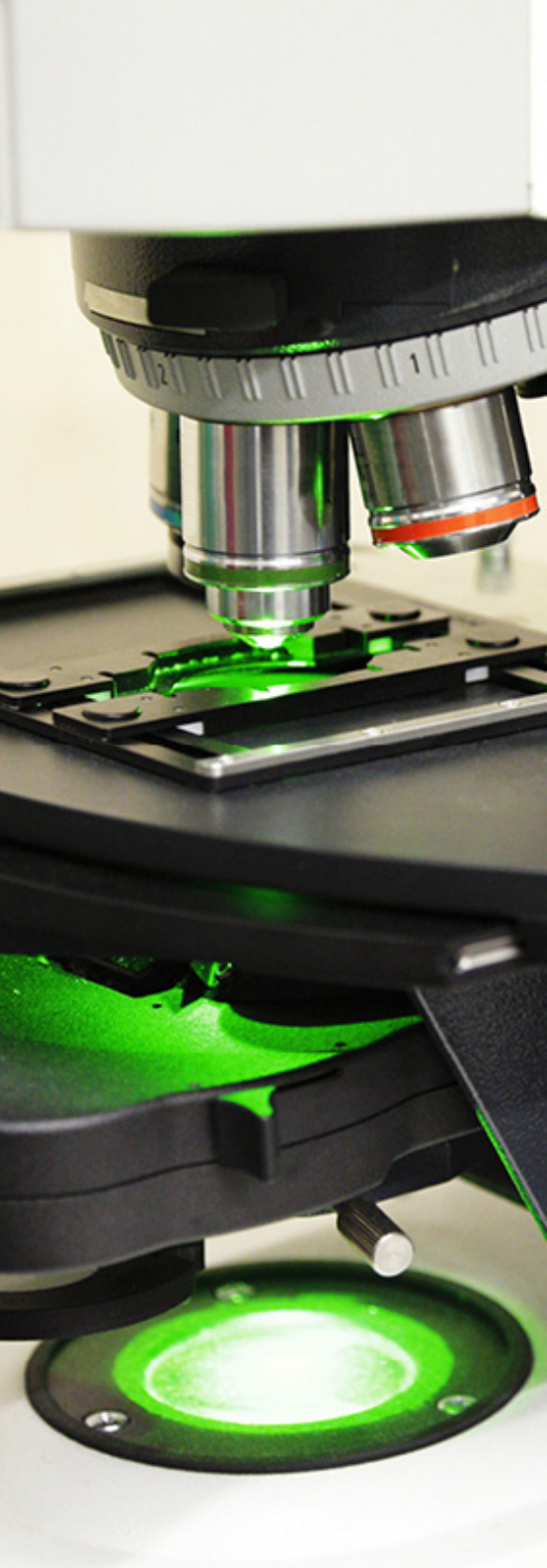
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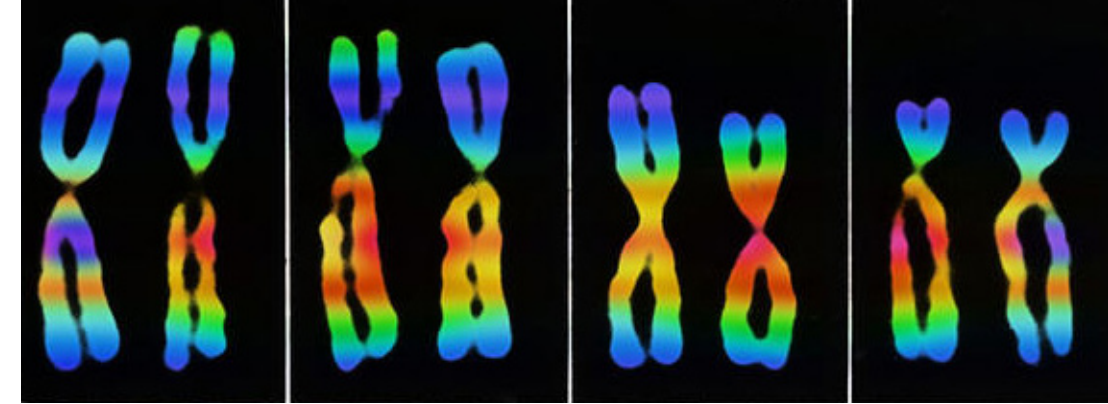
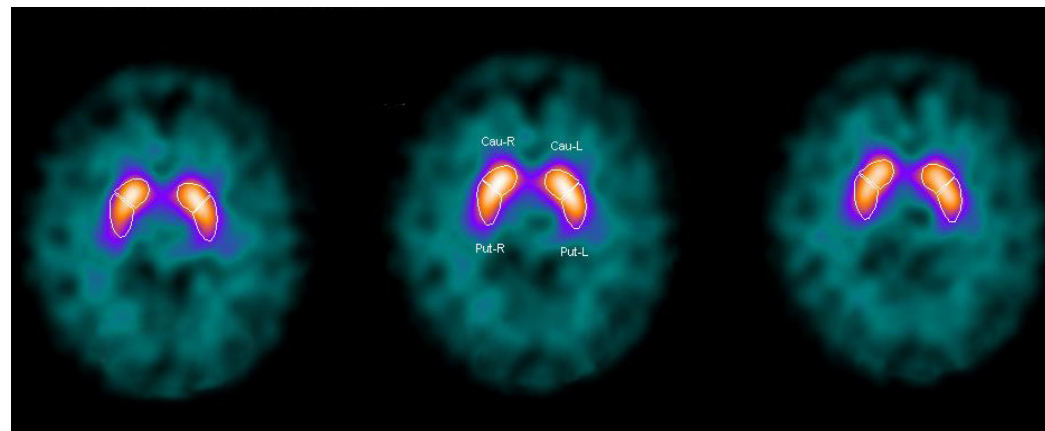
## INSTITUTE OF NEUROLOGICAL SCIENCES

The Institute of Neurological Sciences (ISN) has a main location in Mangone (CS) and two research units in Roccella di Borgia (Catanzaro) and Catania. Its current staff includes 45 employees and 49 post-docs. The primary mission of the ISN is to advance the understanding of neurological disorders by means of a high-quality and interdisciplinary approach. The ISN is a center of excellence where clinical services and research activities thrive together. Its interdisciplinary research program spans across five major scientific fields: neuroimaging, genetics, pharmacology, functional genomics and neurobiology. The Institute incorporates a center for advanced diagnostics that is directly accredited with the Italian healthcare system and offers advanced clinical services to patients. An extensive biological, phenotypic and genotypic repository that currently counts more than 12000 DNA samples and 8000 cell lines provides an invaluable source for a wide array of follow-up studies. Core facilities in biochemistry, genetics, genomics, micro-

scopy, neuroimaging, pharmacology and proteomics offer a multidisciplinary approach and a research environment, which can efficiently deal with the current demands of integration of basic and clinical neurological sciences. In addition to fostering innovative research and spreading of scientific knowledge, the ISN develops advanced diagnostic testing and cutting-edge biotechnologies.

## TECHNOLOGY TRANSFER

Thanks to its expertise in the genomic sector, the ISN research unit of Catania is actively involved in technology transfer and has intense collaborations with private sector companies, participating in the Sicily Technologic District Micro and Nano systems, and in the National Technology Cluster “Alisei” for Life Sciences. In these projects, the ISN is involved in the development and validation of innovative diagnostic biosensors for qualitative and quantitative genetic analysis.



## TRANSLATIONAL NEUROLOGY

A close interaction between basic and clinical neuroscientists at the Institute of Neurological Sciences allows to rapidly translating scientific observations and laboratory discoveries into new approaches for diagnosing, treating, and preventing neurological diseases. An invaluable window on the complexity of human neurological diseases is offered by brain imaging, genetic testing, neuropsychological and biodemographycal analysis. Genomic and proteomic follow up studies, performed on an extensive collection of human samples, contribute to the understanding of the etiology of neurological disorders, most of which are complex or multifactorial. Discoveries are rapidly translated into clinical diagnostic technologies or gene-based tests. An expanding number of these are then offered to patients to improve diagnosis. Concurrently, translational studies are performed on animal and cellular models of diseases to elucidate pathophysiological mechanisms at the molecular, cellular and system levels. Exploitation of disease mechanisms and drug targets raises novel treatment and prevention strategies.

