



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

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| Aceruloplasminemia | Tay Sachs disease |
| Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy | Mitochondrial diseases |
| Ataxias | Neuroacanthocytosis |
| Atrophies | Pantothenate kinase-associated neurodegeneration |
| Neuronal ceroid lipofuscinosis | Neurodegeneration with brain iron accumulation, type 2 |
| Huntington disease | Neurofibromatosis |
| Familial fronto-temporal dementia | Familial amyloid polyneuropathy |
| Dystrophies | Hereditary neuropathy with liability to pressure palsies |
| Epilepsy | Hereditary spastic paraparesis |
| X-linked lissencephaly type 1-SCLH | Amyotrophic lateral sclerosis |
| Alzheimer disease | Rett syndrome |
| Charcot-Marie-Tooth disease | Startle disease |
| Fabry disease | Familial thrombophilia |
| Parkinson disease | |

RADIOLOGY SERVICES

Standard and advanced magnetic resonance imaging

CONTACTS

Mangone

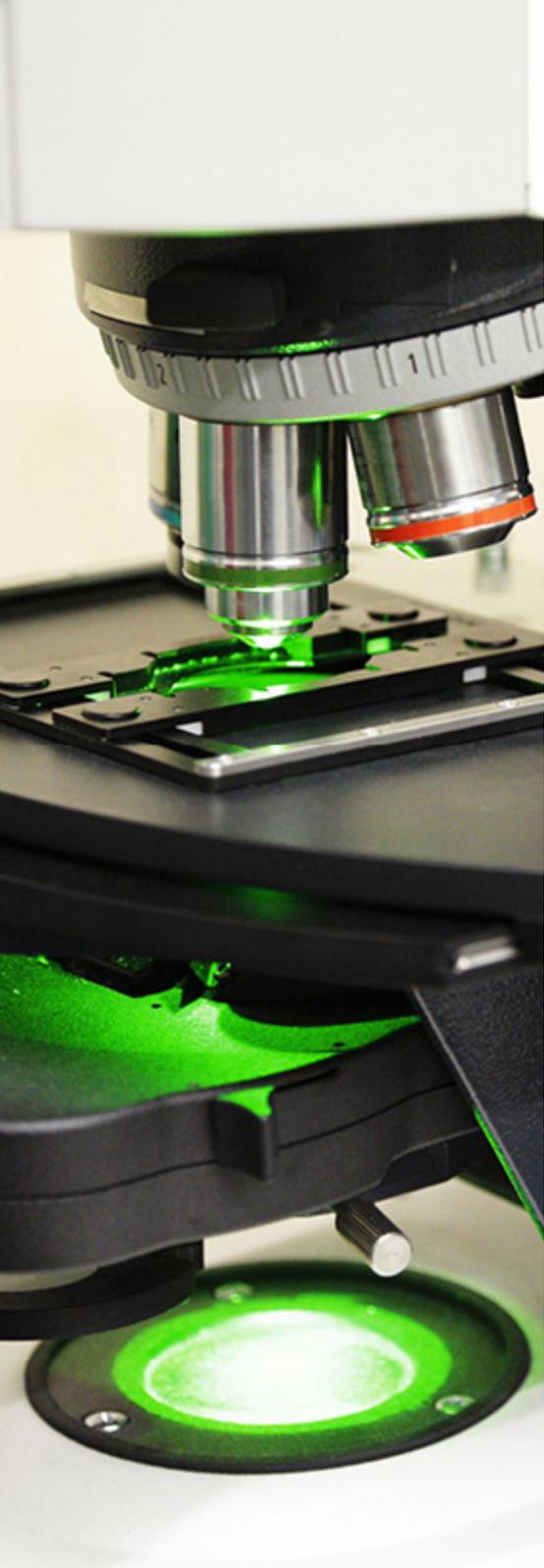
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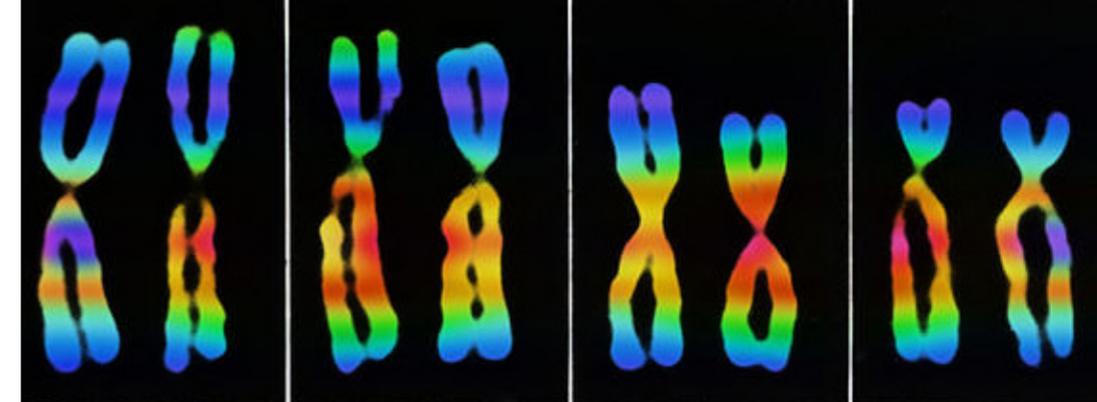
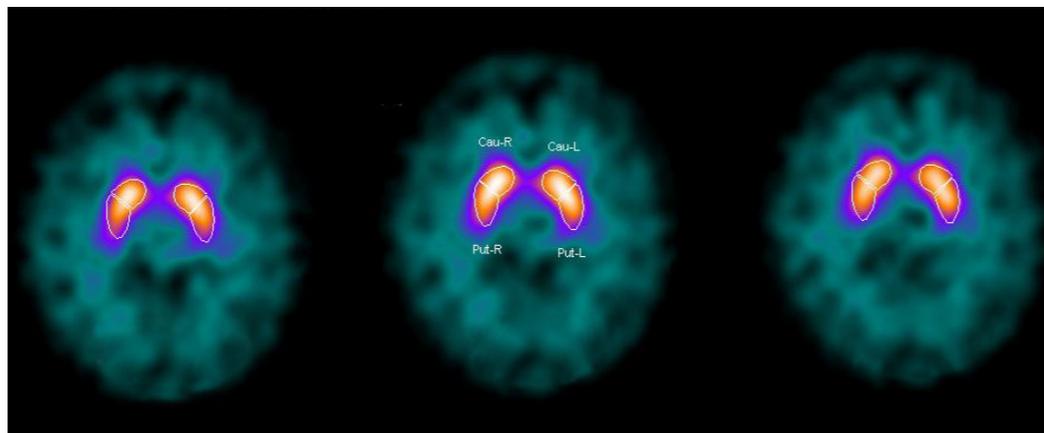
INSTITUTE OF NEUROLOGICAL SCIENCES

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 biodemographical 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.

