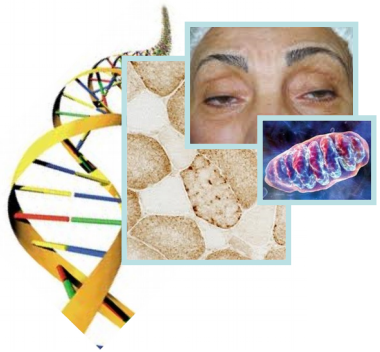

European Conference on Rare Diseases & Orphan Products

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COMBATTI LA DISTROFIA MUSCOLARE E LE ALTRE MALATTIE GENETICHE

The experience of the Nation-wide Italian Collaborative Network of Mitochondrial Diseases

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Introduction:

Mitochondrial disorders (MD) are among the most common genetic disorders (estimated prevalence rate: 5-10/100000) and a major burden for society. However, in contrast to the extraordinary progress in our understanding of the biochemical and molecular bases of MD, we are still extremely limited in our ability to treat these complex and heterogeneous conditions.
Small patient populations represent the major impediment to progress in research and care.

Italian Network of Mitochondrial Diseases

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- Graziella Uziel
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- Liliana Vercelli
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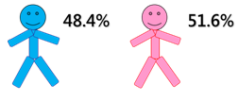
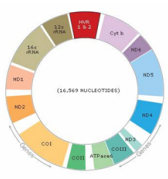


We have collected to date 1289 patients, with both adulthood and childhood onset of the disease.

Molecular diagnosis



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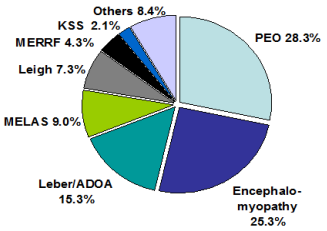
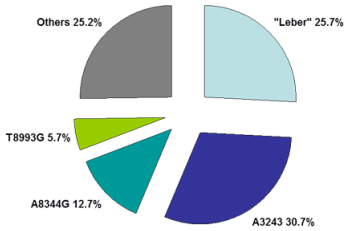
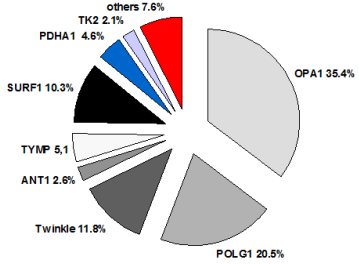
Mitochondrial syndromes



mtDNA point mutations



nuclear point mutations



Material and methods:

We reviewed the features of the carriers of mitochondrial DNA/nuclear mutations enrolled in the web based database of the "Nation-wide Italian Collaborative Network of Mitochondrial Diseases". Eleven centers with expertise on mitochondrial medicine have been involved in this project.

Results:

Up to date, we have collected 1289 patients with both adulthood (52%) and and childhood (48%) onset of the disease. A molecular diagnosis was achieved in 56% of cases (33% mitochondrial DNA, 23% nuclear DNA mutation). The commonest clinical phenotype in our registry is progressive external ophtalmoplegia (28.3%), followed by encephalomyopathy (25.3%), Leber/ADOA (15.3%) and MELAS (9.0%). The most frequent mtDNA mutation were Leber-associated point mutations, A3243G and A8344G while the most frequently mutated nuclear gene were OPA1, POLG1, Twinkle and SURF1.

Conclusions:

MD are rare disease and studies often rely on a few cases. This limitation can be overcome by creating patient registries. The creation of a national register has allowed us to characterize of a big cohort of MD cases with special issue on phenotype genotype correlation and disease natural history, to redefine guidelines and diagnostic criteria for MD.
Large, multicenter studies are strongly needed to better characterize the clinical picture and natural history of these diseases, for the achievement of future clinical trial with potential new drugs for these chronic, still incurable disorders.