# Italian Network of Mitochondrial Diseases

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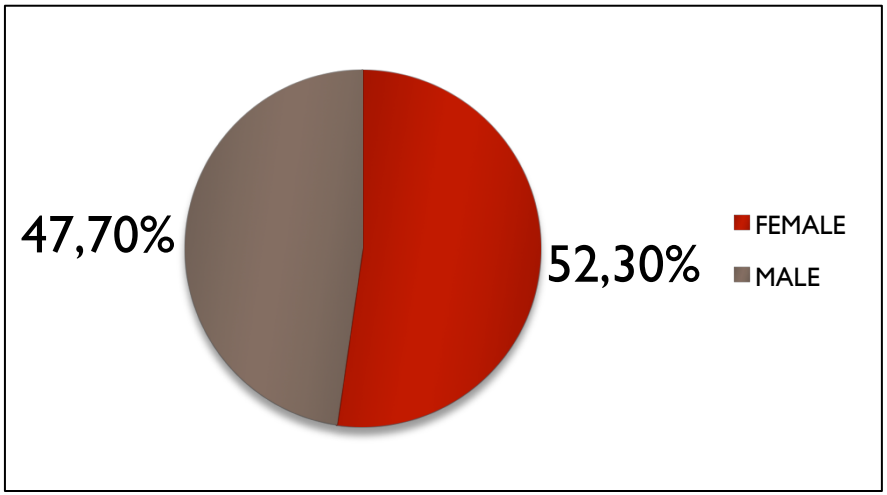
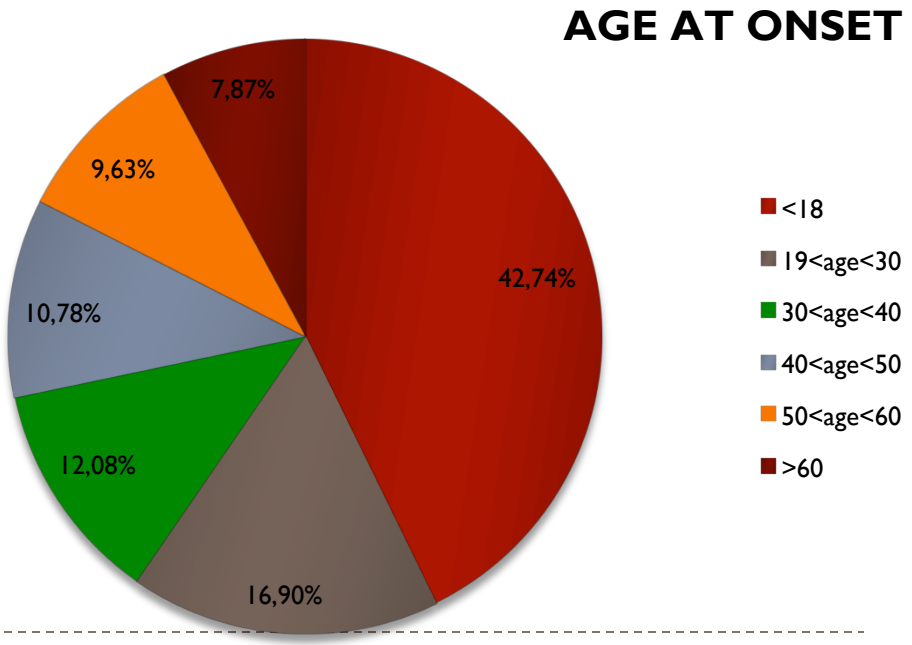
- Filippo Maria Santorelli
- Monica Sciacco
- Serenella Servidei
- Gabriele Siciliano
- Costanza Simoncini
- Paola Tonin
- Antonio Toscano
- Graziella Uziel
- Maria Lucia Valentino
- Liliana Vercelli
- Massimo Zeviani

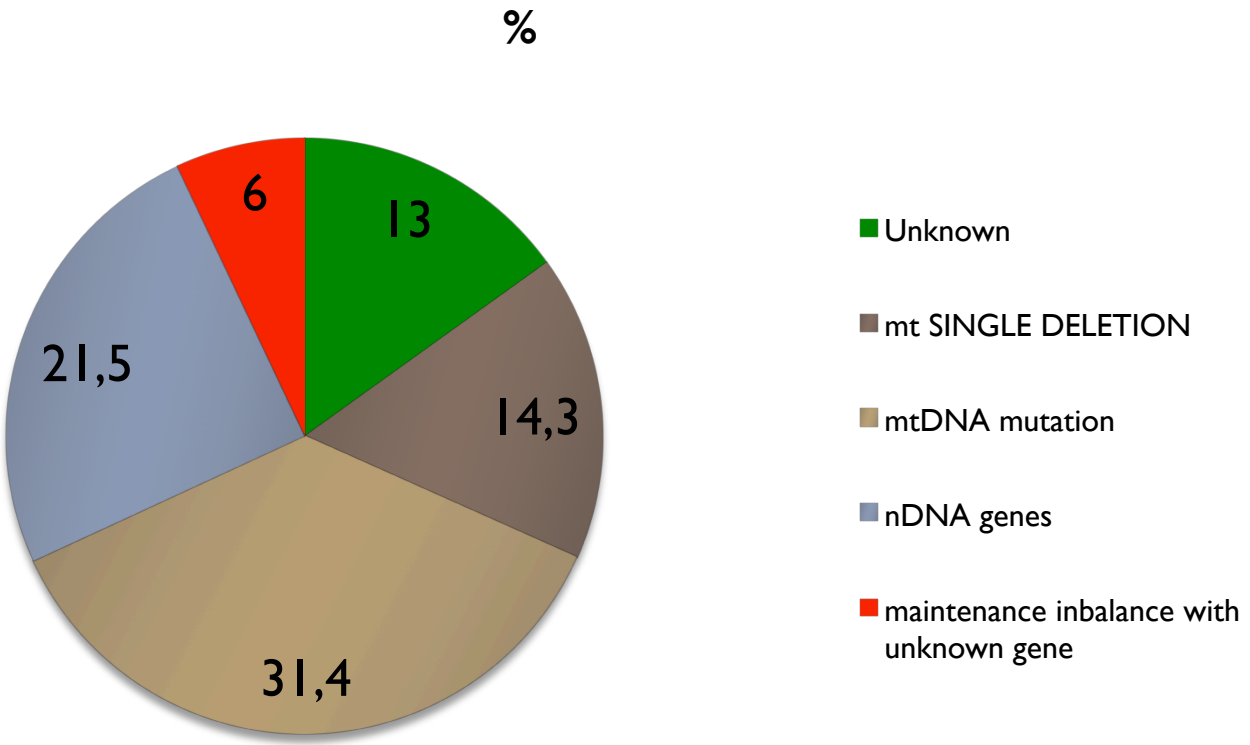
# THE ITALIAN NUMBERS…….

## November 10th, 2018 N= 1753



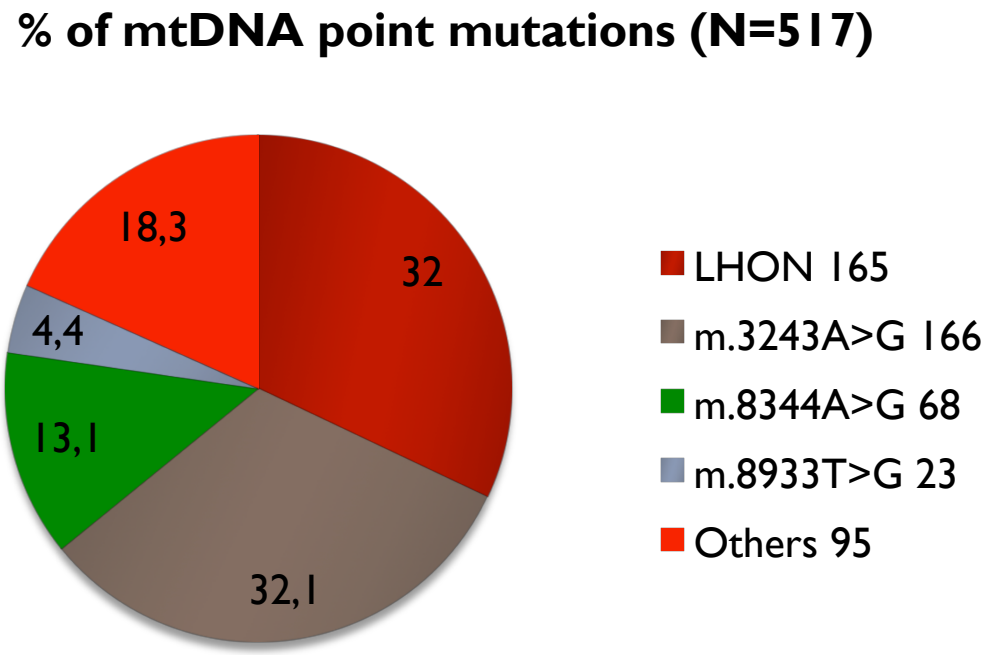



# MOLECULAR ETIOLOGY

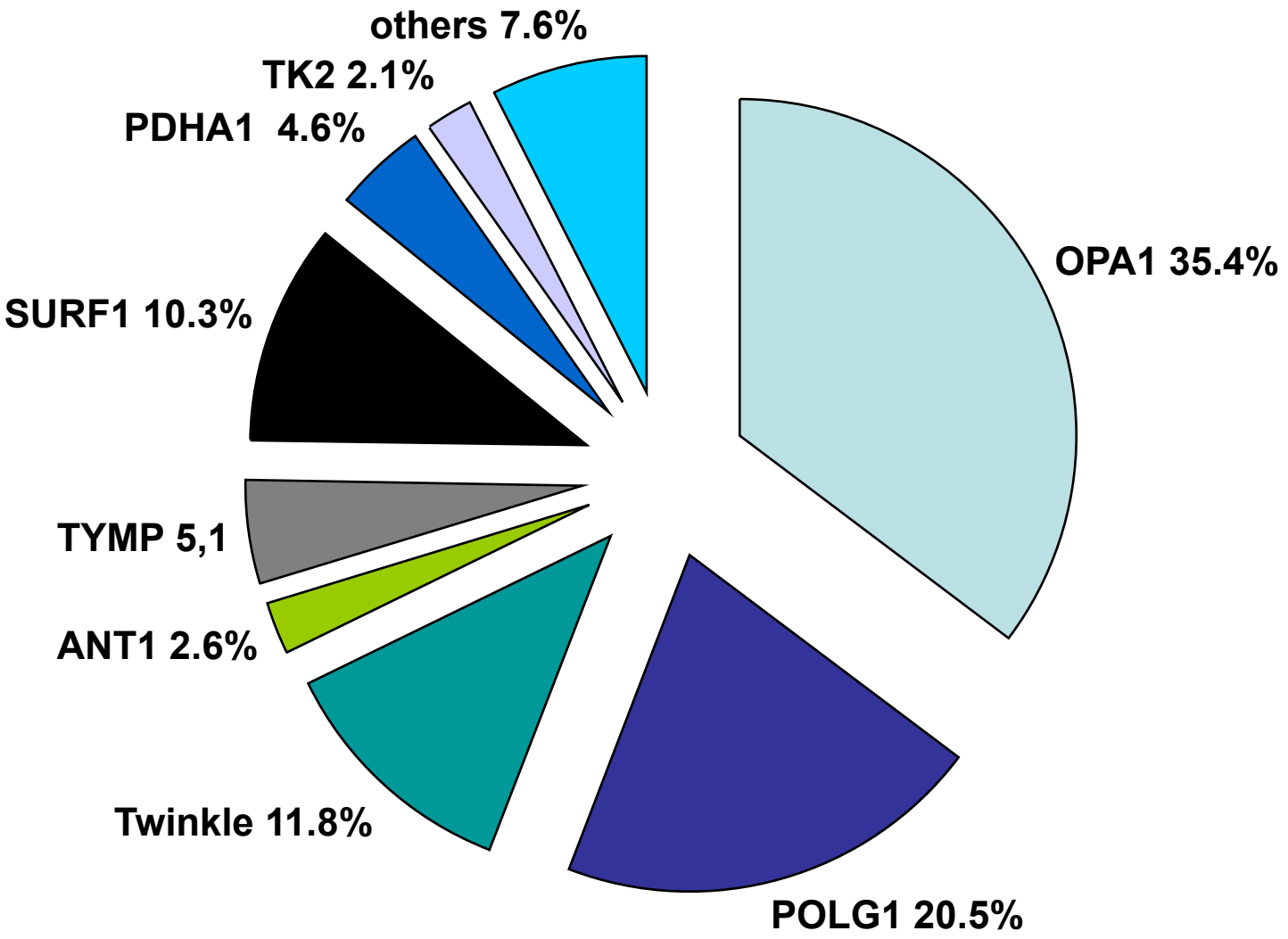


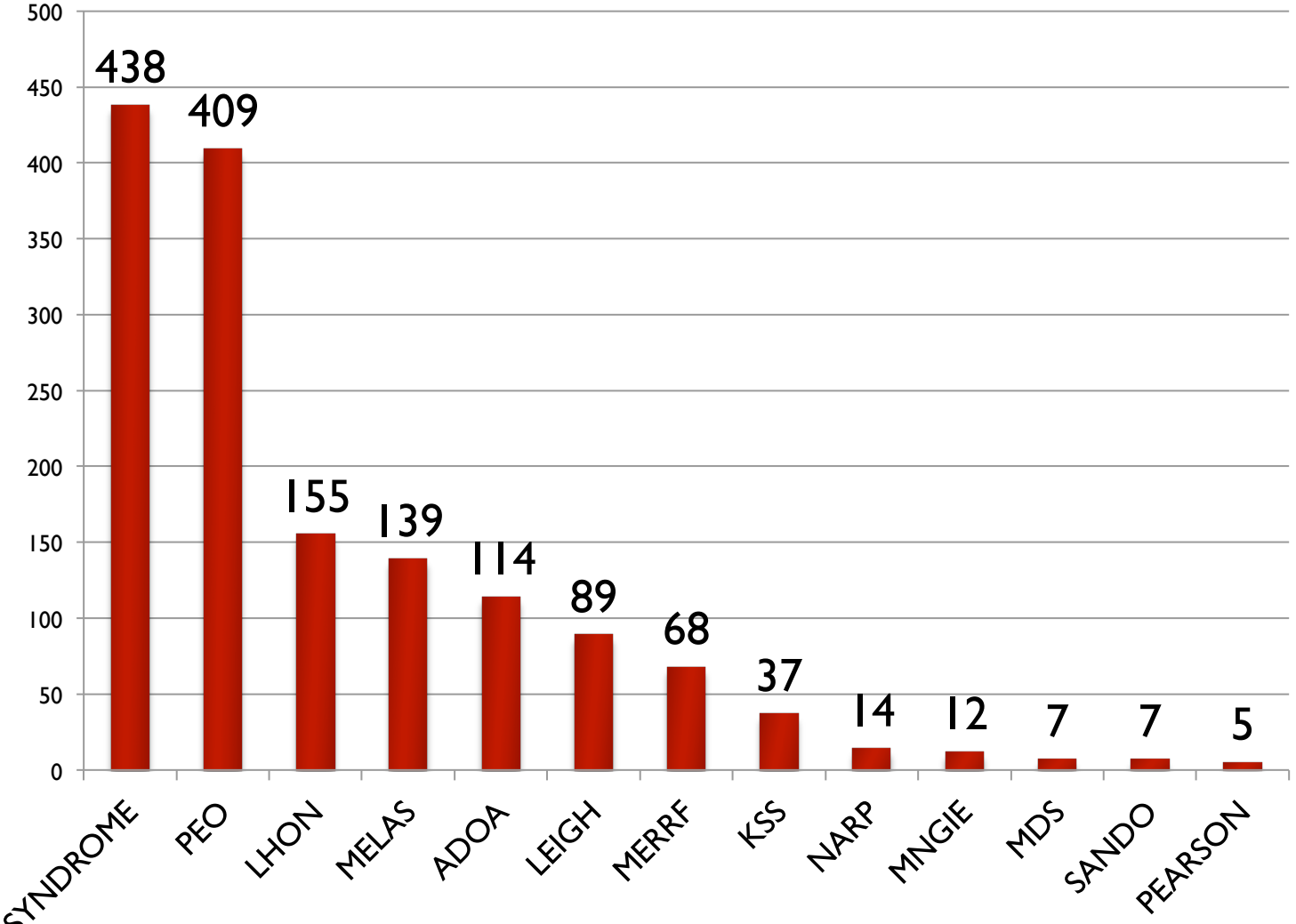

N=1646

# mtDNA point mutations (n=1646pts)



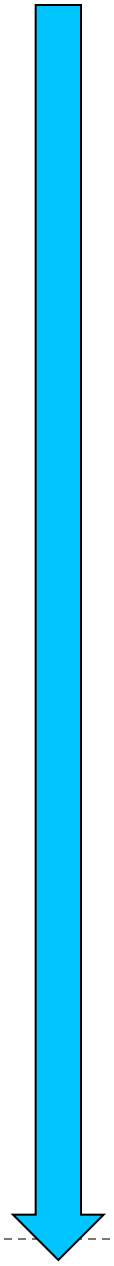
# nuclear point mutations




500
450
400
350
300
250
200
150
100
50
0
438
409
155
139
114
89
68
37
14
12
7
7
5
NO SPECIFIC SYNDROME
PEO
LHON
MELAS
ADOA
LEIGH
MERRF
KSS
NARP
MNGIE
MDS
SANDO
PEARSON

N=1494

| | **Patients** (1330 tot with full phenotype described) | **Percentage** |
|---|---|---|
| **Ptosis/ophthalmoparesis** | 636 | 47,8 |
| **Muscle weakness** | 488 | 36,7 |
| **Hearing loss** | 330 | 24,8 |
| **Exercise intolerance** | 279 | 21 |
| **Optic neuropathy** | 241 | 18,1 |
| **Muscle wasting** | 233 | 17,5 |
| **Cerebellar ataxia** | 198 | 14,8 |
| **Cognitive involvement** | 189 | 14,2 |
| **Hypotonia** | 180 | 13,5 |
| **Neuropathy** | 163 | 12,2 |
| **Swallowing impairment** | 162 | 12,1 |
| **Epileptic seizures** | 152 | 11,4 |
| **Muscle pain** | 152 | 11,4 |
| **Diabetes** | 121 | 9 |
| **Pyramidal involvement** | 115 | 8,6 |
| **Respiratory impairment** | 115 | 8,6 |
| **Cardiomyopathy** | 106 | 7,9 |
| **Migraine** | 102 | 7,6 |
| **Retinopathy** | 92 | 6,9 |
| **Gastroint. dysmotility** | 69 | 5,1 |



J Neurol
DOI 10.1007/s00415-015-7710-y

ORIGINAL COMMUNICATION

## Redefining phenotypes associated with mitochondrial DNA single deletion

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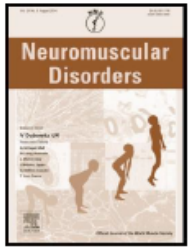
J Neurol
DOI 10.1007/s00415-013-7225-3

ORIGINAL COMMUNICATION

## The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?

Michelangelo Mancuso · Daniele Orsucci · Corrado Angelini · Enrico Bertini · Valerio Carelli · Giacomo Pietro Comi · Alice Donati · Carlo Minetti · Maurizio Moggio · Tiziana Mongini · Serenella Servidei · Paola Tonin · Antonio Toscano · Graziella Uziel · Claudio Bruno · Elena Caldarazzo Ienco · Massimiliano Filosto · Costanza Lamperti · Michela Catteruccia · Isabella Moroni · Olimpia Musumeci · Elena Pegoraro · Dario Ronchi · Filippo Maria Santorelli · Donato Sauchelli · Mauro Scarpelli · Monica Sciacco · Maria Lucia Valentino · Liliana Vercelli · Massimo Zeviani · Gabriele Siciliano

## Accepted Manuscript




Title: "Mitochondrial neuropathies": a survey from the large cohort of the Italian Network

Author: Michelangelo Mancuso, Daniele Orsucci, Corrado Angelini, Enrico Bertini, Valerio Carelli, Giacomo Pietro Comi, Antonio Federico, Carlo Minetti, Maurizio Moggio, Tiziana Mongini, Paola Tonin, Antonio Toscano, Claudio Bruno, Elena Caldarazzo Ienco, Massimiliano Filosto, Costanza

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Neuromuscular Disorders 22 (2012)
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www.elsevier.com/locate/nmd

## Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases

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RESEARCH ARTICLE

## Myoclonus in Mitochondrial Disorders

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## Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation

Michelangelo Mancuso, MD, PhD
Daniele Orsucci, MD
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Enrico Bertini, MD, PhD

ABSTRACT

**Objectives:** Myoclonic epilepsy with ragged-red fibers (MERRF) is a rare mitochondrial syndrome, mostly caused by the 8344A>G mitochondrial DNA mutation. Most of the previous studies have been based on single case/family reports or series with few patients. The primary aim of this study was the characterization of a large cohort of patients with the 8344A>G mutation. The second-ary aim was revision of the previously published data.