

Annual Congress on

Advancements in Neurology, Neuroscience and Pediatric Neurology

June 18-19, 2018 Rome, Italy



Ailian Du

Tongren Hospital-Shanghai Jiaotong University, China

Heteroplasmy of mtDNA 3243A>G mutation in seven Han Chinese families by pyrosequencing

Objective: To study the heteroplasmy and phenotype correlations of mtDNA 3243A>G mutation in seven Han Chinese families using Restrict Fragment Length Polymorphism (RFLP) and Pyrosequencing (Pyro).

Methods: Seven probands were pathologically and genetically diagnosed as mitochondrial diseases with 3243A>G mutation. The clinical phenotypes were studied in 39 maternal family members. 5 were diagnosed as Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-like episodes (MELAS), 2 with pure Mitochondrial Myopathy (MM), 1 with early Neuropathy, Ataxia and Retinitis Pigmentosa (NARP) syndrome. Six with diabetes, 3 with hearing loss and 20 family members are normal. Blood DNA from 37 members were detected with RFLP and pyrosequencing. mtDNA 3243A>G heterogeneity were analyzed.

Results: Mutation load in blood of 5 MELAS patients were 15.7% by RFLP (29% by Pyro), 12.8% (19% by Pyro), 40.1% (53% by Pyro), 25.8% (30% by Pyro), 28.3% (59% by Pyro). Mutation load in 2 MM patients were 13.7% (29% by Pyro) and 76.8% (79% by Pyro), and that in the NARP patient was 20.0% (57% by Pyro). Six family members with diabetes were range from 3.7-7.6% (0-14% by Pyro). Three family members with hearing loss were range from 4-18.2% (6-18%). The mutation load of 14 normal family members ranges from 2% to 12.5% (0-5% by Pyro). Detection by Pyro is more accurate than RFLP when mutation load is lower than 10%. The mutation load is higher those in earlier ages of onset.

Conclusion: Pyrosequencing is more reliable when mutation load is lower than 10%. The mutation load is negatively correlated to the age of onset in this research.

Biography

Ailian Du is the Vice Director of Department of Neurology, Tongren Hospital, Shanghai Jiao Tong University School of Medicine. She was graduated from Huashan Hospital, Fudan University and worked at Second Affiliated Hospital, Zhejiang University School of Medicine for 10 years. She has been studied at University of California San Diego as Visiting Scholar for 2 years from 2008 to 2010. She is specialized in neuro-immune diseases and neuromuscular diseases and advanced on the research of myasthenia gravis and mitochondrial disease.

lotusdu@126.com