Author's response to reviews

Title: Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G>A mitochondrial DNA mutation: a case report.

Authors:

Dario Ronchi (darioronchi@tin.it)
Alessandra Cosi (alessandra.cosi@studenti.unimi.it)
Davide Tonduti (davidetondu@hotmail.com)
Simona Orcesi (simona.orcesi@mondino.it)
Andreina Bordoni (andreina.bordoni@unimi.it)
Francesco Fortunato (francesco.fortunato@unimi.it)
Mafalda Rizzuti (mafalda.rizzuti@studenti.unimi.it)
Monica Sciacco (monica.sciacco@policlinico.mi.it)
Martina Collotta (martina.collotta@studenti.unimi.it)
Sophie Cagdas (sophie.cagdas@aopoma.it)
Giuseppe Capovilla (pippo.capovilla@aopoma.it)
Maurizio Moggio (maurizio.moggio@unimi.it)
Angela Berardinelli (angela.berardinelli@mondino.it)
Pierangelo Veggiotti (pierangelo.veggiotti@unipv.it)
Giacomo P. Comi (giacomo.comi@unimi.it)

Version: 2 Date: 25 May 2011

Author's response to reviews: see over
Reviewer 1 (Dr. Ryan)
We thank Dr Ryan for her comments. A native English speaker has revised the manuscript.

Reviewer 2 (Dr. Yiu)
1. The mother of the proband underwent a general clinical assessment and was reported in good health.
2. The last part of the discussion has been modified. In the absence of functional studies the influence of the novel m.14792C>G variant on the phenotype is speculative. Previously reports of m.14459-mutated families suggest the involvement of modifier factors different from mitochondrial DNA sequence.
3. A native English speaker has carefully revised the manuscript.

Minor points
1. The statement on botox has been removed from the manuscript.
2. Columns relative to laboratory findings and muscle biopsy were removed.