Author's response to reviews

Title: Eight previously unidentified mutations found in the OA1 ocular albinism gene

Authors:

Helene Mayeur (mayeur@necker.fr)
Olivier Roche (roche@necker.fr)
Christelle Vetu (vetu@necker.fr)
Carolina Jaliffa (jaliffa@necker.fr)
Dominique Marchant (marchant@necker.fr)
Alex V Levin (alex.levin@sickkids.ca)
Elise HeON (eheon@attglobal.net)
Joanne Sutherland (joanne.sutherland@sickkids.ca)
Helene Dollfus (Helene.DOLLFUS@chru-strasbourg.fr)
Dominique Bonneau (do-bonneau@numericable.fr)
Francis L Munier (francis.munier@ophtal vd.ch)
Daniel F Schorderet (daniel.schorderet@iro vsnet.ch)
Didier Lacombe (lacombd@nsbx.chu-bordeaux.fr)
Edith Said (edith.said@um.edu mt)
Eddy Mezer (emezer@rambam.health.gov.il)
Josseline Kaplan (emezer@rambam.health.gov.il)
Jean-Louis Dufier (dufier@necker.fr)
Cecile Marsac (marsac@necker.fr)
Maurice Menasche (menasche@necker.fr)
Marc M Abitbol (abitbol@necker.fr)

Version: 4  Date: 6 April 2006

Author's response to reviews: see over
Dear Colleagues,

Thank you for your kind and insightful comments on our Manuscript: manuscript ID 565777918966469. These are our answers point by point to each reviewer. All the modifications of the revised manuscript are in RED.

To the Attention of Dr VALERIA MARIGO:

1) In this version of the manuscript, we looked for deletions in the 27 patients where no mutation was found.

2) I am myself an MD ophthalmologist specialized in France in the study of Albinisms as well as a PhD. There are cases where the mother has very subtle signs and is a carrier of an OA1 mutation and cases where the oculo-cutaneous albinism is mild with blond hairs and a fundus showing a very mid pigmentation. Only 90% of the mothers of children affected by X linked ocular albinism display fundal signs. In 10% of the cases there is nothing found.

3) We agree with Dr MARIGO on the point concerning the THREONINE 166: We introduced the related modifications which suggest also that the mutation of this amino acid residue is pathogenic.

4) Concerning the Frameshift mutation that was potentially susceptible to modify the interaction with the ligand, we modified the sentence according to the suggestion of Dr VARIGO.

5) We included the modifications suggested by Dr MARIGO in the text of the Manuscript.
To The Attention of Dr MARIA-VITTORIA SCHIAFFINO

1) We agree with Dr MARIA-VITTORIA SCHIAFFINO on the point concerning the THREONINE 166: We introduced the related modifications which suggest also that the mutation of this amino acid residue is pathogenic.

2) We provided the information concerning the chromatograms by specifying who was a carrier and who was an affected patient.

3) We modified the text of the manuscript according to the suggestions of Dr SCHIAFFINO.

We were unable to further characterize the deletions or exon skipping due to a shortage in money and reagents.

With our kindest regards

Marc Abitbol MD,PhD