Author's response to reviews

Title: Congenital aplasia of the optic chiasm and oesophageal atresia: a case report

Authors:

Stefano Pensiero (pensiero@burlo.trieste.it)
Paolo Cecchini (paolo.ceccini@libero.it)
Paola Michieletto (paolamic@inwind.it)
Gloria Pelizzo (pelizzo@burlo.trieste.it)
Maurizio Madonia (madoct76@libero.it)
Fulvio Parentin (parentin@burlo.trieste.it)

Version: 2 Date: 11 April 2011

Author's response to reviews: see over
1. We added a table with the clinical characteristics of the syndromic oesophageal atresia and compared them our case. The more frequent are VACTERL syndrome, Feingold syndrome, AEG syndrome and CHARGE syndrome. In the text we added other more frequent causes of OA (chromosomal, maternal diabetes and phenylketonuria).

2. We have correct a little the English language.

3. The white and the black arrow on the MR images point out respectively the presence of the hypoplastic anterior optic nerves and the aplasia of the optic chiasm.

4. As we had described in the text, we did not considered the syndrome correlated to chromosomal defects, because in our case the karyotype was normal, such as we had excluded maternal pathologies, rarely associated with OA (diabetes and phenylketonuria) and any maternal infection or drug abuse during pregnancy. Our patient can not therefore be classified in any of the syndromes associated with OA. Genetic syndromes with OA do not present achiasma, while the genes that determine achiasma not produce syndromes with OA. Genes involved in the described syndromes, as in the updated text, are all on different chromosomes, so it is unlikely their simultaneous impairment. Therefore it seems likely, as VACTERL and OAVS, that this syndrome can be of malformative origine.