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Brief Note

Association of eye and neurological diseases

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The finding of eye abnormalities in patients with neurological disease is important for both clinical management and genetic research (Miller and Newman, 2004). In fact, if we exclude degenerative, vascular and inflammatory causes of both eye and neurological pathologies, in some cases we have a genetic origin of conditions, and it is well known that there is eye involvement in numerous genetic syndromes and diseases characterized by neurological involvement. In a search for "eye and neurological disease" in the OMIM database, we found 27 different conditions (OMIM, 2004).

The prevalence of severe ocular disorders was recently reported in a large-scale study of intellectual disability patients (van Splunder et al., 2004); neurological impairment was a prominent feature.

Recently, we have focused our attention on a possible relationship between heart and ocular defects in Down's syndrome (DS) patients (Bianca and Bianca, 2004; Bianca et al., 2004). Based on two different databases, one a prospective clinical follow-up of DS subjects and the other a congenital malformation registry dataset, we found a significant relationship between congenital cataract and atrial septal defects in both DS and non-DS subjects, in whom there was only eye and heart involvement.

On the basis of these preliminary observations, we suggest that susceptibility genes for specific heart and ocular anomalies could be contiguous, or reciprocally influenced. The same genetic hypothesis may be postulated for some neurological diseases, e.g., neurofibromatosis or

Wilson's disease, in which specific patterns of eye anomalies are recurrently associated with the condition, while others are occasionally reported as if protective mechanisms existed.

A study of a large patient sample with specific recurrent associations may contribute to the search for a possible candidate gene involving both eye and neurological conditions. On the other hand, the association found between ocular and neurological disease suggests that a multidisciplinary approach is needed, and all neurological patients, both syndromic and isolated, should be routinely evaluated by an ophthalmologist to exclude or to early diagnose an ocular condition. This could help prevent permanent visual dysfunction in patients who have severe diseases.

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