Fluctuations in contrast sensitivity and color vision of Leber's hereditary optic neuropathy carriers

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LHON is caused by a mitochondrial DNA mutation that leads to sudden irreversible loss of central vision. Visual alterations may be present in asymptomatic carriers. We report a longitudinal study measuring color vision and contrast sensitivity (CS) in LHON carriers. **Methods.** Forty four LHON carriers were tested yearly (2003-2008) and their thresholds were compared to norms. Color discrimination was assessed along the protan, deutan and tritan axes with the Cambridge Color Test (CCT). Luminance CS was measured with a checkerboard stimulus at two spatial frequencies (0.5 and 4.5 cpd), presented at 33 and 1500 ms. **Results.** Color discrimination was impaired in 47% of the carriers in 2003: 39% in the protan axis, 37% in the deutan and 14%, in the tritan. Over successive years, thresholds changed in 25/44 subjects, improving to normality in 25%, becoming abnormal in 16% and fluctuating from normal to abnormal in 16%. Spearman correlations of protan thresholds, compared year by year, and the same for deutan and tritan, were close to zero in 7 of 12 comparisons. For CS, in the comparison over successive years, thresholds changed in 52% of subjects, having improved to normality in 25%, became abnormal in 11% and fluctuated between normal and abnormal in 17%. Spearman correlation coefficients for CS thresholds were close to zero in the comparison between the three successive years for all measurement conditions. **Conclusion** There are large fluctuations of visual performance in over half of the LHON carriers, possibly caused by changes due to environmental stresses. We believe that these changes could be associated with structural compensations (nerve fiber layer swelling and microangiopathy). If the compensatory structural changes are inadequate or environmental stresses excessive, the LHON carrier may develop the severe optic neuropathy associated with LHON affected patients.

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