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LECTURE

Close encounter on chromosome 14

Sergiu C. Blumen[1,2], Itzhak Braverman[1,2], and Tamar Ben-Yosef[2]

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Abstract

Since the discovery of the large cluster of Oculopharyngeal Muscular Dystrophy (OPMD) among Uzbek (Bukhara) Jews, we noticed that some OPMD homozygotes had significantly reduced vision due to pigmentary retinopathy. Recent investigations in a newly identified OPMD homozygote, as well as in several Uzbek OPMD heterozygotes, revealed in this population, a high prevalence of a novel, autosomal recessive, mutation producing, adult onset, progressive visual loss in homozygotes; vision is not affected in heterozygote carriers. This mutation, occurring on chromosome 14q, in close proximity to PABPN1, was found neither among Uzbeks with two normal [(GCN)10] PABPN1 alleles nor in OPMD patients belonging to another ethnic group. We conclude that, during history, two founder mutations occurred on 14q in Uzbek Jews and they are closely linked.

This research was motivated by Fernando Tome's personal example and dedication to OPMD patients. Each clinical observation must be explained and, if possible, elucidated at molecular level. That is the lesson, the authors learned from him.

[1]The Hillel Yaffe Medical Center, Hadera

[2]Rappaport Faculty of Medicine, The Technion, Haifa, Israel

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