Close encounter on chromosome 14

Sergiu C. Blumen1,2, Itzhak Braverman1,2, and Tamar Ben-Yosef2

1The Hillel Yaffe Medical Center, Hadera
2Rappaport Faculty of Medicine, The Technion, Haifa, Israel

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.