Reply to: [Factor XI mutation and the origin of Ashkenazi Jews. Haematologica 2008; 93:e59]

Our current[1] and previous studies[2] unequivocally showed that the haplotype for factor XI type II mutation observed in Italians is identical with the founder haplotype discerned in Ashkenazi and Iraqi Jews as well as in Arabs.[3] An age estimate disclosed that this ancestral type II mutation appeared in Jews more than 2,500 years ago.[4] These findings make it very likely that type II mutation diffused into the Italian population and the Palestinian Arab population at later times when Jews settled in Italy as from the second century B.C, and after Arabs settled in Israel – in 622 A.D, respectively. Regarding type III mutation, haplotype analysis performed in Italians[2]again identified complete identity with the founder haplotype discerned in Ashkenazi Jews.[3] This mutation has not been observed among 1343 non-Ashkenazi Jews nor among 313 Arabs, and an age estimate revealed that it appeared in Ashkenazi Jews more recently than the type II mutation.[3,4] Whether or not it first occurred in Roman Jews who resided in Rome from the second century B.C is questionable although in 3 out of 107 Roman Jews recently examined, heterozygosity for the type III mutation was detected.[5]

A linkage disequilibrium (LD) analysis, which could be of interest to compare the extent of LD between Italians and Ashkenazi Jews, was not feasible due to the limited number of Italian individuals bearing the type II (sixteen) and/or the type III (eight) mutations.

Zoosmann–Diskin's suggestion that the origin of Ashkenazi Jews in general, and the type III mutation in particular is in the Italian population is untenable because a recent analysis of more than 4000 samples from four genome-wide association studies clearly distinguished among Northwestern Europeans, Southeastern Europeans (including Italians) and Ashkenazi Jews.[6]

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