

CORRIGENDUM

Is the novel SCKL3 at 14q23 the predominant Seckel locus?

Kılınc MO, Ninis VN, Uğur SA, Tüysüz B, Seven M, Balcı S, Goodship J and Tolun A

European Journal of Human Genetics (2015) **23**, 140; doi:10.1038/ejhg.2014.258

Correction to: *European Journal of Human Genetics* (2003) **11**, 851–857; doi:10.1038/sj.ejhg.5201057

The authors report that the results for Seckel Syndrome 3 locus (Kilinc *et al*: *EJHG* 2003; **11**: 851–857; OMIM 608664) were obtained by using superseded genotyping technology, and unfortunately it has become apparent that the results are incorrect.

The reason for the incorrect results is that microsatellite genome scan, which was commonly utilised for disease gene mapping a decade ago, is not adequate in mapping studies in small inbred families. In this study, a low-density microsatellite set containing 156 autosomal markers was used. For one of the families a single locus with a maximal multipoint LOD score >3 was detected, and that locus was published as SCKL3. However, recently the gene defect responsible for the disorder in that family was found in *LIG4* at another locus. *LIG4* is responsible for LIG4 syndrome (features including immunodeficiency and developmental and growth delay), associated with microcephaly as does the Seckel syndrome. Apparently, parental consanguinity was

higher than the authors had assumed; that is why there were >1 loci with identical-by-descent homozygous haplotypes in the patients of the family with moderate size. The smaller, actual disease locus escaped identification with linkage analysis based on microsatellite genotype data.

Since the authors began using SNP genotypes for linkage mapping they frequently detected >1 locus yielding LOD scores >3 in inbred families, as generally they were not aware of all parental blood relationships. In their recent publication on recessive azoospermia genes (Ayhan *et al*: *J Med Genet* 2014; **51**: 239–244), the disease mutation in the four affected brothers was in the 0.23-Mb locus and not in the other possible disease locus of 14-Mb; both loci yielded the same maximal LOD score of 3.01. An analysis based on microsatellite genome scan would have detected only the larger locus, as was in the case for their study on the SCKL3 family.

The authors sincerely regret having inadvertently published an incorrect result. The wrong result was simply due to the limitations of the technology employed, which was acceptable at the time.