Abstract

Gaucher disease is an autosomal recessive disorder caused by the deficiency of β-glucocerebrosidase. Some Gaucher patients carry in their β-glucocerebrosidase genes complex mutations which apparently arose by a recombination with the non-functional β-glucocerebrosidase pseudogene. Recombination between genes and their corresponding pseudogenes plays a role in the development of other hereditary human diseases. Mutant alleles formed in male and female meiosis are a source of these variations in the gene pool. The study of frequency and scope of recombination events in human disease-associated genes in the gametes is of importance for evaluation of the disease burden in the population. The evaluation of the scope of single recombination events in the β-glucocerebrosidase gene in human gametes is technically challenging. Novel technologies such as next-generation sequencing, nanopore sequencing or droplet digital PCR may have advantages over previously used techniques in this application.

Key words: recombination, gene conversion, pseudogene, β-glucocerebrosidase, complex alleles, Gaucher disease