FREQUENCY OF GBA GENE MUTATION N370S, CAUSING GAUCHER DISEASE, IN LATVIA

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BACKGROUND

Gaucher disease is an autosomal recessive disorder of lysosomal degradation of glucosylceramide. It results in a progressive accumulation of glucosylceramide in the lysosomes of macrophages, leading to hepato-splenomegaly, anemia, thrombocytopenia and various skeletal complications. The most common mutation is the substitution A-GG at nucleotide 1226, which produces a Asn-Ser change at amino acid 370. There is variable frequency of this mutation: in Ashkenazic Jews carrier frequency 1: 15; 1 in 100 people in the United States.

AIM

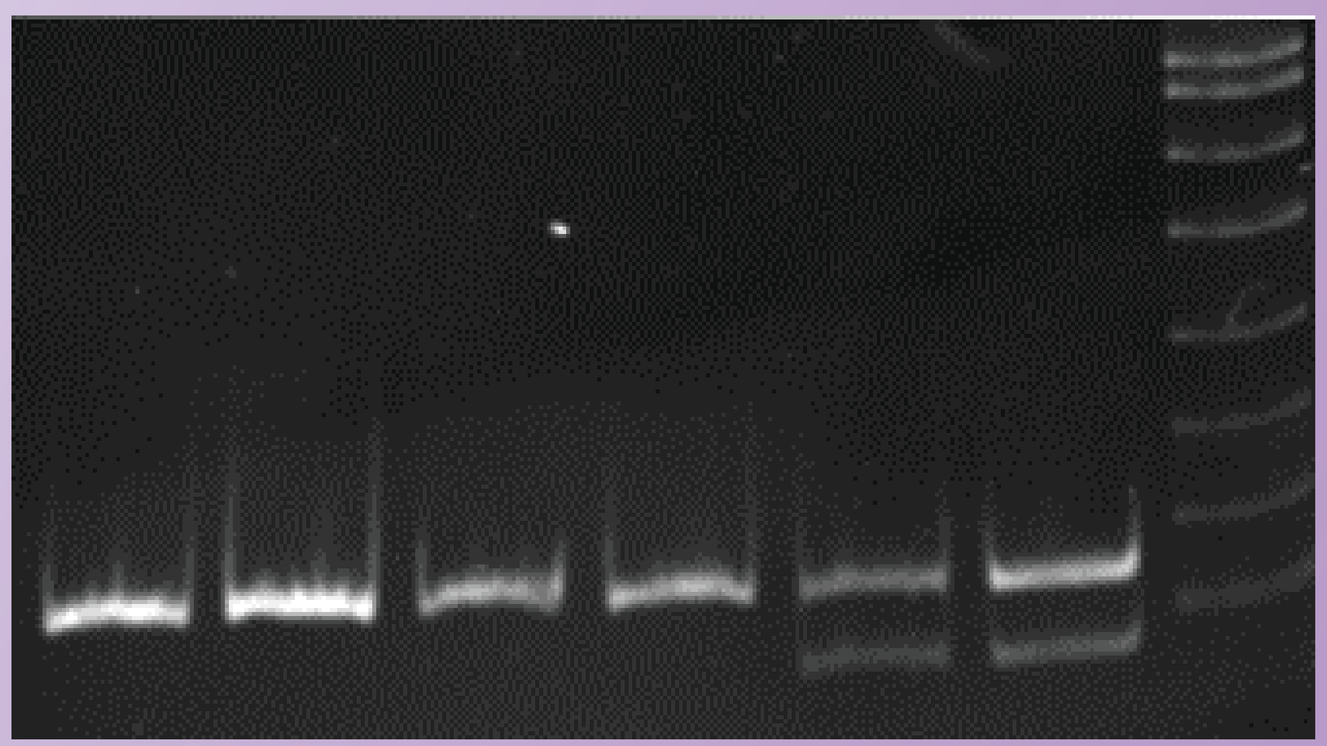
Detect carrier frequency of mutation N370S in Latvian population.

MATERIALS AND METHODS

We studied 150 volunteers, aged 19 – 25 years. DNA was extracted from whole blood and purified by standard phenol/chloroform extraction protocol. The presence of mutation N370S in gene GBA was analyzed using PCR with subsequent restriction enzyme XhoI digestion and detected in polyacrylamide gel.

RESULTS

One patient out of 150 was heterozygous for mutation N370S. Estimated Gaucher disease frequency when patients are homozygous for mutation N370S is 1 : 90 000.



CONCLUSIONS

1. Carrier frequency of mutation N370S in Latvian population is quite low.
2. Gaucher disease in Latvian population may be caused by other mutations.
3. To obtain more precise results about mutation frequency control population should be enlarged.

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