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Therefore, this preliminary finding suggests a disorder in the enzymes or vitamin B12.

In each of the three cases the definitive diagnosis was established by demonstration of impaired cholecalciferol activation in skin fibroblasts. In conclusion, these cases illustrate the diverse, but common presentations of a rare disorder. Pulmonary manifestations (as in case 3) are rarely described in classical NP-C, but these cases illustrate the diagnostic dilemma of NP-C.

In the same report, the authors also analyzed the spectrum of genotypes in Gaucher patients, finding that the N370S/84GG genotype is always associated with a PvuII*/PvuII genotype, but there were exceptions to this rule in some patients. They concluded that the N370S/84GG genotype is associated with a PvuII*/PvuII genotype, and that the N370S/84GG genotype is always associated with a PvuII*/PvuII genotype.

Identification of two novel polymorphisms in the glucocerebrosidase gene region: the first one consists of a tetranucleotide (AAAT) repeat upstream to the glucocerebrosidase gene, and the second is a dinucleotide (CT) repeat in the intergenic region between the glucocerebrosidase gene and its pseudogene. These two polymorphisms, along with the previously reported L444P and 84GG mutations, were studied to determine the spectrum of genetic abnormalities responsible for this disease. The study of these markers may reveal possible ancestral chromosomes which led to affected alleles, leading to the understanding of the origin of glucocerebrosidase mutations.