

## Case Report

# A neurofibromatosis type 1 family report with multiple cases in 3 consecutive Generations

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## Abstract

Neurofibromatosis type 1 (NF1) with the incidence of 1 in 3500 births, is the most common disorder which affects skin and peripheral nervous system. NF1 results from mutations in NF1 gene. The NF1 gene spans 350kbp and to date, nearly 2434 mutations in it were reported. The gene with 100 percent penetrance is located on chromosome 17 encoding neurofibromin protein. Recently, many challenges of its genetic analysis have been overcome through the application of new sequencing techniques. In present study patients with neurofibromatosis type 1 have been characterized from clinical symptoms such as presence of café au lait spot, plexiform neurofibroma, optic nerves involvement, presence of several patients in first degree relatives. These patients were in different ages including 73, 63, 44, 20 with different symptoms and severities of disease. In this communication, a NF1 family with 4 cases in 3 generations has been presented.

**Keywords:** Neurofibromatosis type 1, Neurofibromin, Mutation

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