Case Report

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

Oladnabi M (Ph.D)*1,2, Haddadi T (B.Sc)3, Kianmehr A (Ph.D)1,4 Mansour Samaei N (MD, Ph.D)1,2, Mehri M (M.D)5

1Gorgan Congenital Malformations Research Center, Golestan University of Medical Sciences, Gorgan, Iran. 2Assistant Professor, Department of Medical Genetics, School of Advanced Technologies in Medicine, Golestan University of Medical Sciences, Gorgan, Iran. 3Nurse, School of Nursing and Midwifery, Golestan University of Medical Sciences, Gorgan, Iran. 4Assistant Professor, Department of Medical Biotechnology, School of Advanced Technologies in Medicine, Golestan University of Medical Sciences, Gorgan, Iran. 5Assistant Professor, Department of Internal Medicine, School of Medicine, Golestan University of Medical Sciences, Gorgan, Iran.

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

* Corresponding Author: Oladnabi M (Ph.D), E-mail: oladnabidozin@yahoo.com

Received 8 Feb 2016    Revised 4 Feb 2017    Accepted 4 Feb 2017