Farber disease (lipogranulomatosis) in Iran: Report of 4 novel mutations in acid ceramidase gene (ASAH1 gene)

Fatemeh.Hadipour¹, Zahra.Hadipour¹, Alireza.Tavassoli², Yousef.Shafaghati³

ABSTRACT

Introduction: Farber lipogranulomatosis is a rare inherited condition involving the breakdown and use of fats in the body (lipid metabolism). Characteristics are early-onset subcutaneous nodules, painful and progressively deformed joints, and hoarseness by laryngeal involvement. In affected individuals, lipids accumulate abnormally in cells and tissues throughout the body, particularly around the joints. Three classic signs occur in Farber lipogranulomatosis: a hoarse voice or a weak cry, small lumps of fat under the skin and in other tissues (lipogranulomas), and swollen and painful joints. Affected individuals may also have difficulty breathing, an enlarged liver and spleen (hepatosplenomegaly), and developmental delay.

Case presentation: Here we report 6 patients with the diagnosis of farberlipogranulomatosis in the past 7 years, confirmed by mutation analysis. Clinical pictures in our patient were mostly joint swelling and tenderness, and weak cry, 5 of the patients were female and only one affected boy who died at Newborn period.

Conclusion: Three of them showed hepatosplenomegaly. Three of the patients are alive. We analysed ASAH1 gene and detected 4 novel mutations on them.

Keywords: Farber disease, Lipogranulomatosis, ASAH1 gene, Novel mutation from Iran

1-MD, Sarem Cell Research center, Department of Medical Genetic, Sarem Hospital, Tehran, Iran.
2-MD, Sub Specialty in Pediatric neurology, Markaz Tebi Hospital, Tehran Medical university.
3- MD, Pediatrics and clinical genetics, Sarem cell Research center, Sarem Hospital, Tehran, Iran.

*Corresponding Author, Email: dr.yshafagh@gmail.com