Sjögren - Larsson syndrome caused by novel compound homozygous mutations in ALDH3A2 gene: A case report

Le Tran Anh Ngan[1], Nguyen Le Trung Hieu[1,2], Dang Hoang Hung[1,2], Trinh Ngo Binh[3]

Department of Neurology, Children's Hospital 2, Ho Chi Minh City[1]

University of Medicine and Pharmacy at Ho Chi Minh City, Ho Chi Minh City[2]

Department of Dermatology, Ho Chi Minh City Hospital of Dermato-Venereology, Ho Chi Minh City[3]

ABSTRACT

Background: Sjögren-Larsson syndrome is a rare autosomal recessive neurocutaneous disorder, characterized by the triad of congenital ichthyosis, spastic diplegia or tetraplegia, and mental retardation. This syndrome was firstly reported by Sjögren and Larsson in 1957.

Case: We describe the 3-year-old patient admitted to the Neurology department with complaints of delayed development. She was born preterm at 36 weeks of gestation from an uneventful pregnancy. Dryness of the skin was detected after birth. Desquamation and erythroderma were diagnosed of psoriasis and treated with topical emollients. Her mother reported a delay of motor development in her childhood, but no similar skin disorder was detected. She had febrile seizures six times when she was in one through two years of age. Neurological examination revealed brisk deep tendon reflexes, increased tone, bilateral extensor plantar responses on lower limbs. She was able to speak only 1 word at a time and her vocabulary was limited to 3 words. Her head circumference was within normal range. Dermatological examination revealed generalized scaly ichthyotic lesions predominantly seen on the lower extremities, neck, trunk and buttock, while the central face is spared Ophthalmological examination did not show any abnormality. Magnetic resonance imaging of the brain revealed normal gross anatomy of the brain, with periventricular white matter abnormalities hyperintense on T2-weighted images. Myelination is generally slightly delayed. Other tests including electroencephalogram, metabolic findings, musculoskeletal evaluation were nornal. She was clinically diagnosed with Sjogren Larrson syndrome. To clarify the cause of this syndrome, we investigated mutations of the ALDH3A2 gene by NextSeq Illumina (USA). We detected a frame shift mutation of the ALDH3A2 gene, on chromosome 17:19657842, NM_000382.3:c.779del (NP_000373.1:p.Lys260ArgfsTer6). This mutation confirmed the diagnosis of Sjögren-Larsson syndrome.

Conclusion: We report a case of Sjögren-Larsson syndrome resulting from a novel compound homozygous mutation, and the new mutation has yet been report in ALDH3A2 database. This finding broadens our understanding of the ALDH3A2 gene mutation profile. This can benefit further researchs into the etiology and treatment of Sjögren-Larsson syndrome.