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ILNEB Syndrome

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The syndrome consisting of Interstitial lung disease, Nephrotic syndrome and Epidermolysis bulllosa is considered to be associated with Integrin α3 (ITGA3) gene mutations. To date only six patients are reported: all carried homozygous ITGA3 mutations and presented a dramatically severe phenotype leading to death before age 2 years, from multi-organ failure due to interstitial lung disease & congenital nephrotic syndrome.

A 9-year-old female child second born of third degree consanguinuous marriage presented with complaints of breathlessness, cough and cold intermittent in nature for the past 6 months. The child was apparently normal until 1 year of age.History elicited revealed uneventful antenatal period, ATT undertaken on positive mantoux with development of skin lesions, passing of foamy, frothy urine & teary eyes for the past 5 years. On examination, retarded growth in terms of height & weight with normal mental age and skin lesions along with periorbital edema were noted. Investigations revealed Pulmonary fibrosis suggestive of Interstitial lung disease, Massive proteinuria indicative of Nephrotic syndrome, Grade III Renal parenchymal disease, Epidermolysis bullosa dystrophica. Genetic testing revealed ITGA 3 mutations and a diagnosis of ILNEB Syndrome was reached from the same.

The Patient was administered supportive treatment with necessary antibiotics and other drugs and advised of danger signs and symptoms such as elevated breathlessness, oliguria, renal failure and worsening of general condition. The patient and her family were counselled under the guidance of a qualified psychiatrist and geneticist.

We describe a variant of the extremely rare ILNEB Syndrome carrying unreported missense mutations in the ITGA3 gene, which is responsible for Interstitial Lung disease, congenital Nephrotic syndrome and Epidermolysis Bullosa (ILNEB). Our case is all the more unique in that, of the six ILNEB patients so far reported, who all died within the age of 19 months from multi-organ failure; our case overcame childhood and is now 9 years old and manifests a mild clinical phenotype due to the lack of homozygous mutations as seen in the other cases.

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