J Clin Exp Dermatol Res 2018, Volume 9
DOI: 10.4172/2155-9554-C1-069

17TH EUROPEAN DERMATOLOGY CONGRESS

March 01-03, 2018 | Paris, France

Gaining insights into FAM111B: A recently identified gene implicated in multisystemic fibrosis

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Mutations in the *FAM111B* gene are associated with hereditary fibrosing poikiloderma (HFP), a recently described multisystemic fibrotic disease. We identified three missense mutations (Tyr621Asp, Arg626Gly and Ser628Asn) that span four families of different ethnic origin. However, the physiological and pathological role of FAM111B and mutated forms respectively is yet to be understood. Thus, to provide insights into the molecular function of the gene product of *FAM111B*, we investigated the subcellular localization in cells expressing FAM 111B. Through bioinformatics studies, we identified putative nuclear localization signal (NLS) in the amino acid sequence of *FAM111B*. We then expressed enhanced green fluorescent fusion proteins of wild-type FAM111B (*FAM111B WT)*, mutants derived from HFP patients and truncated constructs (*FAM111B*-M255 and A375, up- and downstream of putative NLS sequence, respectively) in HEK293 cells. The cellular expression and subcellular localization of FAM111B were determined by confocal microscopy and supported by western blot analysis of the subcellular fractions of cells expressing *FAM111B.* We observed a predominantly nuclear expression of FAM111B WT while a diffused (nuclear and cytoplasmic) pattern of expression was seen with *FAM111B-A*375 and patient-derived mutants. These findings suggest that the predicted NLS signal sequence may be active and that the identified mutations resulted in the aberrant translocation of *FAM111B* to the cytoplasm, thus leading to the reported disease phenotype

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