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LETTER TO THE EDITOR

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"Next generation sequencing identifies mutations in GNPTG gene as a cause of familial form of scleroderma-like disease"

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Dear Editor,

With great interest, we read Zrhidri et al.'s paper [1] which reports compound heterozygous mutations in exon 4 and 9 of the GNPTG gene, in a familial scleroderma-like disease. This novel finding represents an important addition to the family of genetic mutations previously associated with multisystemic fibrosis and scleroderma-like diseases in literature. Further, elucidating pathogenetic mechanisms of genetic systemic fibrosis could potentially lead to discovery of effective treatment of auto-immune systemic sclerosis and related diseases, and alleviate severe morbidity and mortality.

Although the authors focused on scleroderma-like manifestations found in Mucolipidosis type III (pseudo-Hurler polydystrophy), it would have also been useful to mention other genes associated with a scleroderma-like phenotype in their study discussion. Some examples are listed below:

- FAM 111B gene for scleroderma and multisystemic fibrosis-like hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP) [2]
- RECQL4 gene for Rothmund Thomson Syndrome (RTS) [3],
- WRN gene for Werner syndrome (WS) [4]
- LMNA gene in Hutchinson-Gilford progeria syndrome (HGPS) [5]

Finally, considering the multifactorial etiology of fibrosis, it would be interesting to see how the new gene (GNPTG) compares to other genes involved in scleroderma-like diseases such as FAM 111B (POIKTMP), RECLQL4 (RTS), WRN (WS) and LMNA (HGPS); and if there are possible gene interactions, considering the similarities in the phenotype produced.

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