J Pharmacogenomics Pharmacoproteomics 2017, 8:4 (Suppl)
DOI: 10.4172/2153-0645-C1-019

Clinical and molecular analysis of patients with Morquio Syndrome and GM1 gangliosidosis

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Morquio A syndrome and GM1 gangliosidosis are lysosomal storage diseases caused by deficiencies of N-acetylgalactosamine-6-sulfatase (GALNS) and β-galactosidase (β-GAL) respectively, due to the mutations in the genes coding for these enzymes. To further characterize the phenotype and the mutation spectrum for these conditions, we analyzed the clinical profiles of Indian patients with these conditions and identified the mutations in the *GALNS* and *GLB1* genes. All the exons and the adjacent intronic sequences of *GALNS* and *GLB1* were amplified and sequenced. We performed *in silico* and structural analyses of novel mutations identified in this study. We identified 57 different mutations in *GALNS* in 110 families, of which 31 were novel and 26 were previously reported. We also found 42 different mutations in *GLB1* in 62 families, of which 26 were novel and 16 were already reported. We identified the most common mutations in *GALNS* and *GLB1*, and noted a higher frequency of mutations in exons 1, 8 and 7 of *GALNS* and exons 1, 14 and 10 of *GLB1*. We propose a diagnostic strategy to identify the mutations in *GALNS* and *GLB1* in Indian patients with these conditions. *In silico* and structural analyses of *GALNS* and *GLB1* revealed that all novel mutations affect the function and structure of the proteins. This study presents the largest series of patients with Morquio A syndrome and GM1 gangliosidosis and is the first from India. This study has helped prenatal diagnosis and genetic counselling of affected families.

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