

SP1-binding site polymorphism in the COL1A1 gene and its relation to osteoporosis in Egyptian patients with Gaucher disease

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Gaucher disease (GD) is a recessive disorder due to a mutation of the glucocerebrosidase gene leading to reduction in β -glucocerebrosidase activity, accumulation of glucosylceramide. Enzyme replacement is the most common therapy; however, bone manifestations can be slow to respond. COL1A1 is one of the principal candidate genes for fragility fractures in osteoporosis. This study examined the distribution of the polymorphic variant in the regulatory region of COL1A1 gene at a recognition site for transcription factor in Egyptian patients with Gaucher Disease compared to a control group. Thirty Egyptian patients with GD from About El Riche Hospital, Cairo University and 30 healthy age and sex-matched individuals from Fayoum University Hospitals were included as controls. Clinical examination was done for both patients and controls. SP1 binding site polymorphic variant at COL1A1 gene was detected by PCR-RFLP for both groups. Over sixty six percent (66.6%) of the GD patients had the wild genotype (G/G) while 26.7% were heterozygous for G/T polymorphism. Only 6.7% harbored the homozygous T/T

variant. The findings make the hypothesis of an association between Sp1 COL1A1 gene polymorphism and bone disease in GD probably feasible and that component strongly influence bone remodeling.

Key words: COL1A1 gene, Gaucher disease (GD), Bone Mineral Density (BMD), Osteoporosis, osteopenia.

Biography

Shahira Elshafie had completed her MD Degree of Clinical and Chemical Pathology in Kasr El-Aini Medical School Cairo University, 1996. She had received the Basic Certificate of Medical Education from Cairo University in 2016. She is the head of Department of Clinical and Chemical Pathology, Fayoum University from 2016. She had received the Best Paper Award, Initial Steps for Internationalization of Medical Education in Egypt and International Business and Education Conference, Las Vegas, 2014. She is the Egyptian Syndicate of Medical Doctors, since 1984 and Founding member of the Women in Higher Education for Leadership, 2011.

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