Abstract: The human GLB1 gene encodes a lysosomal \(\beta\)-galactosidase (\(\beta\)-Gal) and an elastin-binding protein (EBP). Defect of the EBP as a chaperon for tropoelastin and a component of receptor complex among neuraminidase-1 (NEU1) and protective protein/cathepsin A (PPCA) is suggested responsible for impaired elastogenesis in autosomal recessive \(\beta\)-Gal, PPCA and NEU1 deficiencies. The purpose of this study is to determine effects of GLB1, PPCA and NEU1 gene mutations on elastogenesis in skin fibroblasts. Elastic fiber formation and the EBP mRNA expression were examined by immunofluorescence with an anti-tropoelastin antibody and RT-PCR selective for EBP in skin fibroblasts with these lysosomal enzyme deficiencies. Apparently normal elastogenesis and EBP mRNA expression were observed for fibroblasts from Morquio B disease cases with the GLB1 gene alleles (W273L/W273L, W273L/R482H and W273L/W509C substitutions, respectively), a galactosialidosis case with the PPCA allele (IVS7+3A/IVS7+3A) and a sialidosis case with the NEU1 allele (V217M/G243R) as well as normal subject. In this study, the W273L substitution in the EBP could impossibly cause the proposed defect of elastogenesis, and the typical PPCA splicing mutation and the V217M/G243R substitutions in the NEU1 might hardly have effects on elastic fiber formation in the dermal fibroblasts. J. Med. Invest. 53 : 103-112, February, 2006

Keywords: elastin-binding protein, lysosomal \(\beta\)-galactosidase gene, lysosomal enzyme deficiencies, morquio B disease, costello syndrome
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Elastogenesis in lysosomal β-galactosidase, cathepsinA and neuraminidase-1 deficiencies
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a) β-Gal cDNA
EBP cDNA
F592 F430 F435 F390 F351 F451 F319 F622

β-Gal
EBP
β-Actin

GM1-Gangliosidosis Morquio B

b) F642 F350 RT

EBP
β-Actin
(a) EBP mRNA

(b) \(\beta\)-Gal mRNA
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