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Genomic structure of human GM3 synthase gene (hST3Gal V) and identification of mRNA isoforms in the 5-untranslated region

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GM3 synthase, which transfers CMP-NeuAc with an α 2,3-linkage to a galactose residue of lactosylceramide, plays a key role in the biosynthesis of all complex gangliosides. In this study, cDNA and genomic clones encoding human GM3 synthase (hST3Gal V) were isolated, and the structural organization of the gene was determined. The hST3Gal V cDNA was identical in the coding region with cDNA that has been cloned previously from the HL-60 cells but different in the 5'-untranslated region. The hST3Gal V gene consisted of 9 exons, which span approximately 44 kb, with exons ranging in size from 112 to 1242 bp. The coding region was located in exons 4-9, and all exon-intron boundaries except the acceptor site of intron 1 followed the GT-AG rule. The expression of this gene was highly restricted in both human fetal and adult tissues. By comparison of the nucleotide sequences of the genomic DNA with cDNA sequences including 5'-RACE products, we identified four isoforms (type 1-4) of the hST3Gal V mRNA that differ only in the 5'-UTR. analysis of these isoforms suggests that mRNA isoforms of hST3Gal V are produced by a combination of alternative splicing and alternative promoter utilization.