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Introduction to Bioinformatics
Bioinformatics Analysis of BTN1A1

My chosen sequence, ACC number NM_001732.3, is a nucleotide sequence. This sequence is found in the human genome (National Center for Biotechnology Information).

Gene mapping history: “Accession NM_001732 was first seen at NCBI on Mar 24, 1999 05:08 PM” (National Center for Biotechnology Information). The sequence was updated to its current version (v. 3) on Jul 11, 2019, and the flatfile has had minor updates as recent as Apr 29, 2025.

Length: NM_001732.3 is 2945 bp in length (National Center for Biotechnology Information).

Associated proteins: For accession number NM_001732.3, the encoded protein is NP_001723.2 (Butyrophilin precursor). This transcript corresponds to the gene BTN1A1 (butyrophilin subfamily 1 member A1) in *Homo sapiens*. The mRNA sequence maps to chromosome 6 on the human genome (GRCh38 assembly: NC_000006.12, positions 26,500,303–26,510,425; T2T-CHM13 assembly: NC_060930.1, positions 26,368,710–26,378,831). It is also cross-referenced as CCDS4614.1 and Ensembl gene ENSG00000124557. The related sequences include the mRNA (NM_001732.3), the protein (NP_001723.2), and the genomic sequence (NC_000006.12/NC_060930.1).

Protein coding: The BTN1A1 sequence is unique because it encodes butyrophilin, the major protein of the milk fat globule membrane, essential for milk fat secretion in lactating mammary glands. It belongs to the immunoglobulin superfamily and is located within the MHC class I region, linking it to immune system genes. Recent research also highlights BTN1A1 as a novel immune checkpoint, giving it a dual role in nutrition and immunity.

Flatfile references:

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Entrez

When searching Entrez Gene, I found that BTN1A1 shows a number of single nucleotide polymorphisms (SNPs) scattered across its gene sequence, including coding SNPs, missense SNPs, and non-coding SNPs. These can affect codon usage, protein interactions, and gene regulation, respectively. In addition to SNPs, BTN1A1 has some small insertions or deletions reported in non-coding regions. These can alter promoter or enhancer sequences and may make small changes to the amount of gene expression. BTN1A1 also has multiple mRNA isoforms, created by alternative splicing. These isoforms usually encode the same protein but may differ in regulatory regions that are not translated. The presence of multiple isoforms gives cells flexibility in how the gene is expressed under different conditions, such as during lactation. Entrez Gene also shows that BTN1A1 has conserved orthologs in other mammals, including humans and cattle, reflecting its important role in milk fat secretion across species.

Ensembl

According to Ensembl Gene, BTN1A1 has “2 splice variants, 1 gene allele, 98 orthologs, and 15 paralogs” (Ensembl). These versions code for proteins with the same important domains, but they differ in length (BTN1A1-204 has 2945 base pairs, and BTN1A1-203 has 2895 base pairs) and in regions that don’t directly code for protein, which could change how the gene is regulated. Ensembl also showed several SNPs in the gene. Some of these are located in coding regions where they could alter an amino acid, while others are in non-coding regions that might affect expression levels. Ensembl also shows that BTN1A1 has orthologs in other mammals like cattle and mice. Overall, the database highlights how the gene is both conserved and variable, depending on the region being looked at.

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