



Protein Designing Studies and Biophysical Characterization of Gaucher Disease

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Received: 30 Jan 2019 / Accepted: 20 Feb 2019 / Published online: 01 Apr 2019

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Abstract

Gaucher disease a series of disorders that is due to deficient activity of the enzyme glucocerebrosidase, which leads to accumulation of glucocerebroside in tissues of the body. The five types of Gaucher disease encompass a continuum of clinical findings from a lethal form that occurs before or just after birth to a form so mild that it may not be diagnosed until old age. All types of Gaucher disease are inherited in an autosomal recessive manner. The *GBA* gene provides instructions for making an enzyme called beta-glucocerebrosidase. This enzyme is active in lysosomes, which are structures inside cells that act as recycling centers. Lysosomes use digestive enzymes to break down toxic substances, digest bacteria that invade the cell, and recycle worn-out cell components. Glucocerebroside is a component of the membrane that surrounds cells. It gets broken down by beta-glucocerebrosidase when cells die, and the components are reused as new cells are formed. The sequence of glucosidase beta acid (GBA) retrieved from National Centre for Biotechnology Information in fasta format. The sequence analysis of glucosidase beta acid (GBA) were carried out by using bioinformatics tools like Protparam, Radar, Smart tools and structural designing of glucosidase beta acid were carried out by using CPH server. Further studies are required to investigate the glucosidase beta acid (GBA) of for potential pharmacological properties.

Keywords

Gaucher disease, *GBA* gene, Protparam, Radar, Smart, CPH server.

INTRODUCTION

Gaucher disease (GD) encompasses a continuum of clinical findings from a perinatal lethal disorder to an asymptomatic type. The identification of three major clinical types (1, 2, and 3) and two other subtypes (perinatal-lethal and cardiovascular) is useful in determining prognosis and management.

GD type 1 is characterized by the presence of clinical or radiographic evidence of bone disease (osteopenia, focal lytic or sclerotic lesions, and osteonecrosis), hepatosplenomegaly, anemia and thrombocytopenia, lung disease, and the absence of primary central nervous system disease.

GD types 2 and 3 are characterized by the presence of primary neurologic disease; in the past, they were distinguished by age of onset and rate of disease progression, but these distinctions are not absolute.

- Disease with onset before age two years, limited psychomotor development, and a rapidly progressive course with death by age two to four years is classified as GD type 2.
- Individuals with GD type 3 may have onset before age two years, but often have a more slowly progressive course, with survival into the third or fourth decade.

The *perinatal-lethal form* is associated with ichthyosiform or collodion skin abnormalities or with

nonimmune hydrops fetalis. The *cardiovascular form* is characterized by calcification of the aortic and mitral valves, mild splenomegaly, corneal opacities, and supranuclear ophthalmoplegia. Cardiopulmonary complications have been described with all the clinical subtypes, although varying in frequency and severity.

METHODOLOGY

Target Protein of Glucosidase Beta Acid (GBA) sequence were retrieved from NCBI data base. The retrieved sequences is submitted to the following server and tools, for protein profiling and functional annotation. The retrieved sequence is submitted to the protparam tool and radar tool, for the identification of primary sequence analysis in glucosidase beta acid (GBA). The retrieved sequence is submitted to the SOPMA, Smart, and scan prosite tool for the identification of secondary structural analysis in glucosidase beta acid (GBA). The retrieved protein sequence was applied into CPH server in order to predict 3D dimensional structure of glucosidase beta acid (GBA). The modeled protein 3D structure was viewed with the help of advanced visualization molecular software called Jmol in order to identify the structural region and classify the entire structure of 3D element. The retrieved sequence is submitted to the dipole movement server tool, for the biophysical characterization of glucosidase beta acid (GBA). The retrieved sequence is submitted to the ANNIE tool, for the identification of motif in glucosidase beta acid (GBA). The retrieved sequence is submitted to the CAST-P server, for the identifications of functional units in glucosidase beta acid (GBA).

RESULTS AND DISCUSSION

1. GENE SELECTION:

NCBI:

The gene sequences are collected from using NCBI data base. Among the collected gene sequence. DNA polymerase subunit gamma-1 [Homo sapiens] were selected for further analysis.

A. PROTEIN SEQUENCE:

>NP_001119603.1 DNA polymerase subunit gamma-1 [Homo sapiens]
MSRLLRKVGATVGPVPAPGRWVSSVPASDPSDG
QRRRQQQQQQQQQQQQQPQVLSSEGGQLR
HNPLDIQMLSRGLHEQIFGQGEMPGAAVRRSVEHLQK
HGLWGQPAVPLPDVELRLPLYGDNLQDHF
LLAQKQSLPYLAANLLLQAQLPPKPPAWAWAEGWTRYG
PEGEAVPVAIPEERLALVDFVEVCLAEGTCPT
LAVAISSAWYSWCSQRLVEERYSWTSQSLPADLIPLEVPT
GASSPTQRDWQEQLVGHNVSFDRHIRE
QYLIQGSRMRLDTMSMHMAISGLSSFQRSLWIAAKQK
HKVQPPTKQKQSKRARRGPAISSWDWLDI
SSVNSLAEVHRLVYGGPPLEKEPRELFVKGTMKDIRENFQD
LMQYCAQDVWATHEVFQQQLPLFLERCPH

PVTLAGMLEMGVSYLPVNQNWERYLAEAGQTYEELQREM
KKSMDLANDACQLLSGERYKEDPWLDLEW
DLQEFKQKAKKVKKEPATASKLPIEGAGAPGDPMDQEDL
GPCSEEEFQQDVMARACLQKLKGTTELLP
KRPQHLPHPGWYRKLCPRLLDPAWTPGPSLLSLQMRVT
PKLMALTWDGFLHYSERHGWGYLVPGRRDN
LAKLPTGTTLESAGVVCYRAIESLYRKHCLEQKQQLMPQ
EAGLAEFFLLDNSAIWQTVLEEDYLEVE
AEAKMENLRAAVPGQPLALTARGGPKDTQPSYHHNGPY
NDVDIPGCWFFKLPHKDGNSCNVGSFPAKDF
LPKMEDGTLQAGPGGASGPRALEINKMISFWRNAHKRISS
QMVVWLPRALPRAVIRHPDYDEEGLYAI
LPQVVTAGTITRAVEPTWLTASNARPDRVGSELKAMVQA
PPGYTLVGADVDSQELWIAAVLGDAHFAGM
HGCTAFGWMTLQGRKSRGDLHSTATTVGISREHAKIFN
YGRYAGAGQPAERLLMQFNHRLTQQEAAE
KAQQMYAATKGLRWYRLSDEGEWLRELNLVDRTEGG
WISLQDLRKVQRETARKSQWKKEVVAERAWK
GGTESEMFNKLESIASTDPRTPLVGGCISRALEPSAVQEEF
MTRSVNWVVQSSAVDYLHMLVAMKWL
EEFAIDGRFCISIHDEVRYLVREEDRYRAALALQITNLLTRCM
FAYKLGLNDLPQSVAFFSAVDIDRLR
KEVTMDCKTPSNPTGMERRYGIPQGEALDIYQIELTKGSLE
KRSQPGP

B. NUCLEOTIDE SEQUENCE:

>NM_001126131.1:271-3990 Homo sapiens DNA polymerase gamma, catalytic subunit (POLG), transcript variant 2, mRNA
ATGAGCCGCTGCTCTGGAGGAAGGTGGCCGGCGCCAC
CGTCGGGCCAGGGCCGTTCCAGCTCCGGGGC
GCTGGGTCTCCAGCTCCGTCCTCCGCGCTCCGACCCAGCG
ACGGGCAGCGCGCGCGCAGCAGCAGCAGCA
GCAGCAGCAGCAGCAGCAACAGCAGCCTCAGCAGCCGC
AAGTGCTATCCTCGGAGGGCGGGCAGCTGCGG
CACAACCCATTGGACATCCAGATGCTCTCGAGAGGGCTG
CACGAGCAAATCTTCGGGCAAGGAGGGGAGA
TGCCTGGCGAGGCCGCGGTGCGCCGACGCTCGAGCAC
CTGCAGAAGCACGGGCTCTGGGGGCGAGCCAGC
CGTGCCCTTGCCCGACGTGGAGCTGCGCCTGCCGCCCT
CTACGGGGACAACCTGGACCACTTCCGC
CTCCTGGCCAGAAAGCAGAGCCTGCCCTACCTGGAGGCG
GCCAACTTGCTGTTGCAGGCCAGCTGCCCC
CGAAGCCCCGGCTTGGGCTGGGCGGAGGGCTGGACC
CGGTACGGCCCCGAGGGGAGGCCGTACCCGT
GGCATCCCCGAGGAGCGGGCCCTGGTGTTCGACGTGG
AGGTCTGCTTGGCAGAGGGAAGTTCGCCACACA
TTGGCGGTGGCCATATCCCCCTCGGCTGGTATTCTGGT
GCAGCCAGCGGCTGGTGAAGAGCGTTACT
CTTGACCAAGCAGCTGTGCGCGGCTGACCTCATCCCCCT
GGAGGTCCCTACTGGTGCCAGCAGCCCCAC
CCAGAGAGACTGGCAGGAGCAGTTAGTGGTGGGGCACA
ATGTTTCCTTTGACCGAGCTCATATCAGGGAG
CAGTACCTGATCCAGGGTCCCGCATGCGTTTCTGGACA
CCATGAGCATGCATGGCCATCTCAGGGC
TAAGCAGCTCCAGCGCAGTCTGTGGATAGCAGCCAAGC
AGGGCAAACACAAGGTCCAGCCCCCACAA

GCAAGGCCAGAAAGTCCAGAGGAAAGCCAGAAAGAGGCC
CAGCGATCTCATCTGGGACTGGCTGGACATC
AGCAGTGTCAACAGTCTGGCAGAGGTGCACAGACTTTAT
GTAGGGGGGCTCCCTTAGAGAAGGAGCCTC
GAGAAGTGTGTGAAGGGCACCATGAAGGACATTCGTG
AGAACTCCAGGACCTGATGCAGTACTGTGC
CCAGGACGTGTGGGCCACCCATGAGGTTTTCCAGCAGCA
GCTACCGCTCTTCTGGAGAGGTGTCCCCAC
CCAGTGAAGTGTGGCCGATGCTGGAGATGGGTGTCTCC
TACCTGCCTGTCAACCAGAACTGGGAGCGTT
ACCTGGCAGAGGCACAGGGCACTTATGAGGAGCTCCAG
CGGGAGATGAAGAAGTCGTTGATGGATCTGGC
CAATGATGCCTGCCAGCTGCTCTCAGGAGAGAGGTACAA
AGAAGACCCCTGGCTCTGGGACCTGGAGTGG
GACCTGCAAGAATTAAGCAGAAGAAAGCTAAGAAGGT
GAAGAAGGAACAGCCACAGCCAGCAAGTTGC
CCATCGAGGGGGCTGGGGCCCTGGTATCCCATGGATC
AGGAAGACCTCGGCCCTGCAGTGAGGAGGA
GGAGTTTCAACAAGATGTCATGGCCGCGCTGCTTGA
GAAGCTGAAGGGGACCACAGAGCTCCTGCCC
AAGCGGGCCAGCACCTTCTGGACACCCTGGATGGTAC
CGGAAGCTCTGCCCCGGCTAGACGACCCTG
CATGGACCCCGGGCCAGCCTCCTCAGCCTGCAGATGC
GGGTACACCTAACTCATGGCACTTACCTG
GGATGGCTTCCCTCTGCACTACTCAGAGCGTCATGGCTG
GGGCTACTTGGTGCTGGGCGCGGGACAAC
CTGGCCAAGCTGCCGACAGGTACCACCCTGGAGTCAGCT
GGGGTGGTCTGCCCTACAGAGCCATCGAGT
CCCTGTACAGGAAGCACTGTCTCGAACAGGGGAAGCAG
CAGCTGATGCCCCAGGAGGCCGCTGGCGGA
GGAGTTCCTGCTCACTGACAATAGTGCCATATGGCAAAC
GGTAGAAGAAGTGGATTACTTAGAAGTGGAG
GCTGAGGCCAAGATGGAGAAGTGGAGCTGCAGTGCC
AGGTCAACCCCTAGCTCTGACTGCCCCTGGTG
GCCCCAAGGACACCCAGCCAGCTATCACCATGGCAATG
GACCTTACAACGACGTGGACATCCCTGGCTG
CTGGTTTTTCAAGCTGCCTCACAAGGATGGTAATAGCTGT
AATGTGGGAAGCCCTTTGCCAAGGACTTC
CTGCCAAGATGGAGGATGGCACCTGCAGGCTGGCCC
AGGAGGTGCCAGTGGGCCCCGTGCTCTGAAAA

TCAACAAAATGATTTCTTTCTGGAGGAACGCCATAAAC
GTATCAGCTCCAGATGGTGGTGTGGCTGCC
CAGGTGAGCTCTGCCCCGTGTGTGATCAGGCACCCCGA
CTATGATGAGGAAGGCCTCTATGGGGCCATC
CTGCCCCAAGTGGTACTGCCGGCACCATCACTCGCCGG
GCTGTGGAGCCCACATGGCTCACCAGCCAGCA
ATGCCCCGGCTGACCGAGTAGGCAGTGAGTTGAAAGCC
ATGGTGCAGGCCCCACCTGGCTACACCCTTGT
GGGTGCTGATGTGGACTCCCAAGAGCTGTGGATTGCAGC
TGTGCTTGGAGACGCCACTTTGCCGGCATG
CATGGCTGCACAGCCTTTGGGTGGATGACACTGCAGGGC
AGGAAGAGCAGGGGCACTGATCTACACAGTA
AGACAGCCACTACTGTGGGCATCAGCCGTGAGCATGCCA
AAATCTCAACTACGGCCGCATCTATGGTGC
TGGGCAGCCCTTTGCTGAGCGCTTACTAATGCAGTTTAAAC
CACC GGCTCACACAGCAGGAGGCAGCTGAG
AAGGCCAGCAGATGTACGCTGCCACCAAGGGCCTCCGC
TGGTATCGGCTGTCGGATGAGGGCGAGTGGC
TGGTGAGGGAGTTGAACCTCCAGTGGACAGGACTGAG
GGTGGCTGGATTTCCCTGCAGGATCTGCGCAA
GGTCCAGAGAGAACTGCAAGGAAGTCACAGTGGAAGA
AGTGGGAGGTGGTTGCTGAACGGGCATGGAAG
GGGGGCACAGAGTCAGAAATGTTCAATAAGCTTGAGAG
CATTGCTACGTCTGACATACCACGTACCCCGG
TGCTGGGCTGCTGCATCAGCCGAGCCCTGGAGCCCTCGG
CTGTCCAGGAAGAGTTTATGACCAGCCGTGT
GAATTGGGTGGTACAGAGCTCTGCTGTTGACTACTTACA
CCTCATGCTTGTGGCCATGAAGTGGCTGTTT
GAAGAGTTTGCCATAGATGGGCGCTTCTGCATCAGCATC
CATGACGAGGTTGCTACCTGGTGCGGGAGG
AGGACCGCTACCGCGCTGCCCTGGCCTTGAGATCACCA
ACCTCTTGACCAGGTGCATGTTTGCTTACAA
GCTGGGTCTGAATGACTTGCCCCAGTCAGTCGCCTTTTTT
AGTGAGTCGATATTGACCGGTGCCTCAGG
AAGGAAGTGACCATGGATTGTAAACCCCTTCCAACCCA
ACTGGGATGGAAAGGAGATACGGGATTCCCC
AGGGTGAAGCGCTGGATATTTACCAGATAATTGAAGTCA
CCAAAGGCTCCTTGAAAAACGAAGCCAGCC
TGGACCATAG

The above results show the protein sequence of glucosidase beta acid (GBA).

2. PRIMARY ANALYSIS:

A. PROTPARAM:

<u>10</u> MSRLLRKVA	<u>20</u> GATVGPVP	<u>30</u> APGRWVSSV	<u>40</u> PASDPSDQQR	<u>50</u> RRQQQQQQQQ	<u>60</u> QQQQQPQQPQ
<u>70</u> VLSSEGGQLR	<u>80</u> HNPLDIQMLS	<u>90</u> RGLHEQIFGQ	<u>100</u> GGEMPGEAAV	<u>110</u> RRSVEHLQKH	<u>120</u> GLWGQPAVPL
<u>130</u> PDVELRLPPL	<u>140</u> YGDNLQHFH	<u>150</u> LLAQKQSLPY	<u>160</u> LEAANLLLQA	<u>170</u> QLPPKPPAWA	<u>180</u> WAEGWTRYGP
<u>190</u> EGEAVPVAIP	<u>200</u> EERALVFDVE	<u>210</u> VCLAEGTCPT	<u>220</u> LAVAISSAW	<u>230</u> YSWCSQRLVE	<u>240</u> ERYSWTSQLS

250 PADLIPLEVP	260 TGASSPTQRD	270 WQEQLVVGHN	280 VSFDRAHIRE	290 QYLIQGSRRMR	300 FLDTMSMHMA
310 ISGLSSFQRS	320 LWIAAQGKH	330 KVQPPTKQGQ	340 KSQRKARRGP	350 AISSWDWLDI	360 SSVNSLAEVH
370 RLYVGGPPLE	380 KEPRELFVKG	390 TMKDIRENFQ	400 DLMQYCAQDV	410 WATHEVFQQQ	420 LPLFLERCPH
430 PVTLAGMLEM	440 GVSYLPVNQN	450 WERYLAEAQG	460 TYEELQREMK	470 KSLMDLANDA	480 CQLLSGERYK
490 EDPWLWDLEW	500 DLQEFKQKKA	510 KKVKKEPATA	520 SKLPIEGAGA	530 PGDPMQDQDL	540 GPCSEEEEFQ
550 QDVMARACLQ	560 KLKGTTELLP	570 KRPQHLPQHP	580 GWYRKLCPR	590 DDPAWTPGPS	600 LLSLQMRVTP
610 KLMALTWDGF	620 PLHYSERHGW	630 GYLVPGRRDN	640 LAKLPTGTTL	650 ESAGVVCPYR	660 AIESLYRKHC
670 LEQKGQQLMP	680 QEAGLAEFL	690 LTDNSAIWQT	700 VEELDYLEVE	710 AEAKMENLRA	720 AVPGQPLALT
730 ARGGPKDTQP	740 SYHHGNGPYN	750 DVDIPGCWFF	760 KLPHKDGNSC	770 NVGSPFAKDF	780 LPKMEDGTLQ
790 AGPGGASGPR	800 ALEINKMISF	810 WRNAHKRISS	820 QMVVWLPRSA	830 LPRAVIRHPD	840 YDEEGLYGAI
850 LPQVVTAGTI	860 TRRAVEPTWL	870 TASNARPDV	880 GSELKAMVQA	890 PPGYTLVGAD	900 VDSQELWIAA
910 VLGDAHFAGM	920 HGCTAFGWMT	930 LQGRKSRGTD	940 LHKTATTVG	950 ISREHAKIFN	960 YGRIGAGQP
970 FAERLLMQFN	980 HRLTQQEAAE	990 KAQQMYAATK	1000 GLRWYRLSDE	1010 GEWLVRELNL	1020 PVDRTGGWI
1030 SLQDLRKVQR	1040 ETARKSQWKK	1050 WEVVAERAWK	1060 GGTESEMFNK	1070 LESIATSDIP	1080 RTPVLGCCIS
1090 RALEPSAVQE	1100 EFMTSRVNWV	1110 VQSSAVDYLIH	1120 LMLVAMKWLF	1130 EEFAIDGRFC	1140 ISIHDEVRYL
1150 VREEDRYRAA	1160 LALQITNLLT	1170 RCMFAYKLGL	1180 NDLPQSVAFF	1190 SAVDIDRCLR	1200 KEVTMDCKTP
1210 SNPTGMERRY	1220 GIPQGEALDI YQIELTKGS LEKRSQPGP				1230

The above results shows the primary sequence analysis of glucosidase beta acid(GBA).

B. RADAR :

No. of Repeats	Total Score	Length	Diagonal	BW-From	BW-To	Level
3	328.16	96	484	7	163	1

45- 141 (165.02/125.61) QQQQQQQQQQQQQPQQPQLSSSEGGQ~~R~~HNP~~L~~DIQ~~M~~LS~~R~~GLHEQIFGQ.....GGMPGEAA.VRRSVEHLQ~~K~~HLGQPAV~~L~~PDV~~E~~RLPPLYGDNLDQHFRL
496- 612 (152.82/63.39) KQ~~K~~KAK~~V~~KVKEPATASKLPIEGAGAPGPMQ~~D~~QGLGPCSEEEFFQQdmarac~~l~~q~~k~~lkggtellpk~~r~~PHQLPHG~~P~~WY~~R~~KLCPL~~R~~DDPA.WT.PGPSLSLQ~~M~~RVTPKLMALTNDGFPL
662- 688 (10.32/23.91)EQ.GK.....QQLMPQEA~~G~~.LAEFFLLTNSIAW.

No. of Repeats	Total Score	Length	Diagonal	BW-From	BW-To	Level
2	233.54	75	573	309	410	5

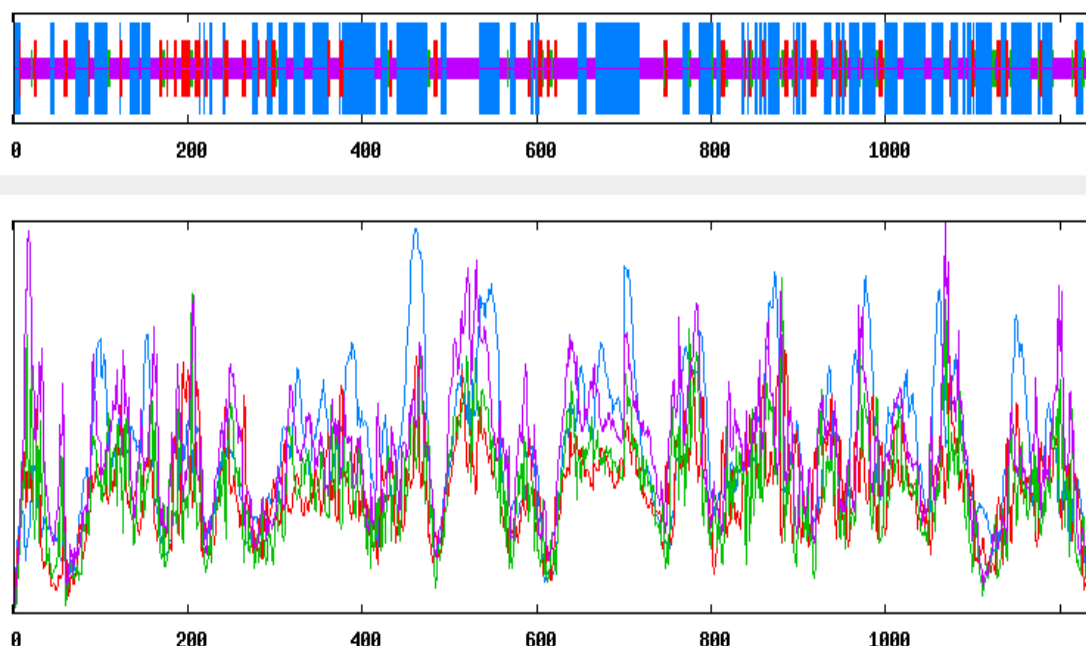
309- 410 (119.39/136.17) **RS**LWIAAKQG.KH..KVQPPTKQGGKS.ORKARRGPAISSIDWLDISSMISLAEVH...RLVVGGPPIekeprelfvgktmkdiren**FQD..L**MQ/caqdvwa**THEV**FOQ
894- 977 (114.16/78.28) **QE**LWIAAVLGdAHfaGIGCTAFGIMTIGRKS**RG**TDLHS(TATTVGISRE**HAK**IFnyg**RI**YGAGQP.....FAEr**L**MOF.....NHRL**TO**QE

The above results show the motif region in glucosidase beta acid (GBA) here hydrophobicity of the amino acids is indicating in different colour.

3. SECONDARY ANALYSIS:

A. SOPMA

[illegible]



Parameters :

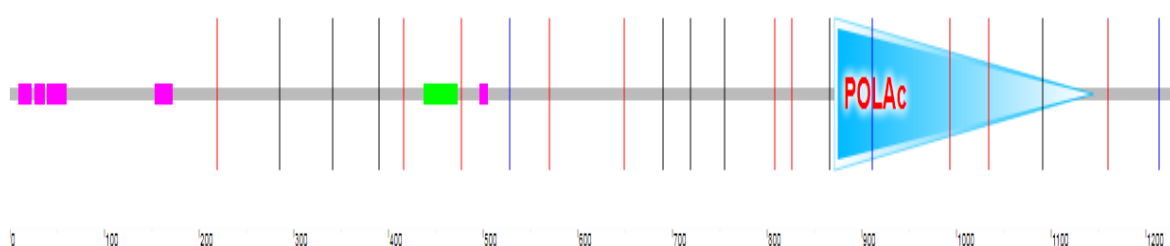
Window width : 17
Similarity threshold : 8
Number of states : 4

The above results shows the secondary sequence analysis of glucosidase beta acid(GBA).

B.SMART:

Domains within *Homo sapiens* protein DPOG1_HUMAN (P54098)

DNA polymerase subunit gamma-1



Information	Architecture	Interactions	PTMs	Orthology
Length	1239 aa			
Source database	UniProt			
Identifiers	DPOG1_HUMAN, P54098, ENSP00000268124, ENSP00000399851, Q8NFM2, Q92515, E5KNU5_HUMAN, E5KNU5, Q2V8X9_HUMAN, Q2V8X9, Q6LCA9_HUMAN, Q6LCA9			
Source gene	ENSG00000140521			
Alternative splicing	DPOG1_HUMAN, HOYD36_HUMAN, HOYD2_HUMAN, HOYDF1_HUMAN, HOYCV2_HUMAN, HOYE43_HUMAN			

Confidently predicted domains, repeats, motifs and features:

Name	Start ▲	End	E-value	
low complexity	9	23	N/A	▲
low complexity	26	37	N/A	
low complexity	39	60	N/A	
low complexity	153	172	N/A	
coiled coil	437	473	N/A	
low complexity	496	505	N/A	
POLAc	871	1145	1.72e-113	▼

Click on a row to highlight the feature in the diagram above. Click the feature name for more information.

The above results show the identification and annotation of genetically mobile domains and the analysis of domain architectures in glucosidase beta acid (GBA). protein.

4.MOTIF SEARCH:

A. SCANEPROSITE:

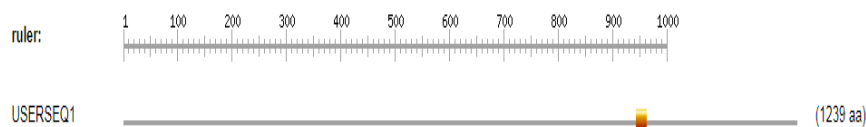
MSRLLWRKVAGATVGPVPAPGRWVSSVSPASDPDQRRRQQQQQQQQQQQQQQPQQPQVLSSEG
GQLRHNPLDIQMLSRGLHEQIFGQGGEMPGEAAVRRSVEHLQKHGLWGQPAVPLPDVELRLPPLYG
DNLDOHFRLLAQKQSLPYLEAANLLQAQLPPKPPAWAWAEGWTRYGPEGEAVPVAIPEERALVFD
VEVCLAEGTCPTLAVAISSAWYSWCSQRLVEERYSWTSQLSPADLIPLEVPTGASSPTQRDWQEQ
LVVGHMVSFDRAHIREQYLIQGSRMRLDTMSMHMAISGLSSFQRS LWIAAKQKGKHKVQPPTKQGG
KSQRKARRGPAISSWDWLDISSVNSLAEVHRLVYGGPPLEKEPRELFVKGTMKDIRENFQDLMQYC
AQDVWATHEVFQQQLPLFLERCPHPVTLAGMLEMGVSYLPVNQNWERYLAEAQGTYEELQREMKS
LMDLANDACQLLSGERYKEDPWLWDLENDLQEFKQKKAKKVKEPATASKLPIEGAGAPGDPMDQE
DLGPCSEEEEFQQDVMARACLQKLKGTTELLPKRPQHLPGHPGWYRKLCPRLLDDPAWTPGPSLLSL
QMRVTPKLMALTWDGFPLHYSERHGWGLVPGRRDNLAKLPTGTTLESAGVVCYPYRAIESLYRKHC
LEQGKQQLMPQEAGLAEFLLTDNSAIWQTVEELDYLEVEAEAKMENLRAAVPGQPLALTARGGPK
DTQPSYHHGNGPYNDVDIPGCWFFKLPHKDGNSCNVGSPPFAKDFLPKMEDGTLQAGPGGASGPRAL
EINKMISFWRNAHKRISSQMWWLPRSAIPRAVIRHPDYDEEGLYGAILPQVVTAGTITRRAVEPT
WLTASNARPDRVGSELKAMVQAPPGYTLVGADVDSQELWIAAVLGDAHFAGMHGCTAFGMMTLQGR
KSRGTDLHSKTATTVGISREHAKIFNYGRIYGAGQPFAERLLMQFNHRLTQQEAAEKAQQMYAATK
GLRWYRLSDEGEWLVRNLNLPVDRTEGGWISLQDLRKVQRETARKSQWKKWEVVAERAWKGGTESE
MFNKLESIAATSDIPRTPVLGCCISRALEPSAVQEEFMTSRVNWVQSSAVDYHLHMLVAMKWL FEE
FAIDGRFCISIHDEVRYLVREEDRYRAALALQITNLLTRCMFAYKLGNDLPQSVAFFSAVDIDRC
LRKEVTMDCKTPSNPTGMERRYGIPOGEALDIYQIIELTGKSLEKRSQGP

Legend:



Please note that the graphical representations of domains displayed hereafter are for illustrative purposes only, and that their colors and shapes are not intended to indicate homology or shared function. For more information about how these graphical representations are constructed, go to <https://prosite.expasy.org/mydomains/>.

hits by patterns: [1 hit (by 1 pattern) on 1 sequence]



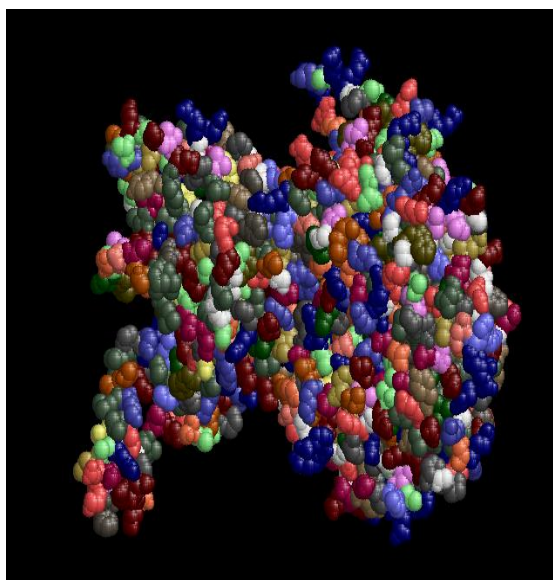
PS00447 DNA_POLYMERASE_A DNA polymerase family A signature :

943 - 962: [confidence level: (0)] RehAKifnYGriYgaGopfA

horizontal scaling:
do not show text labels: ☐
do not show sites in hits: ☐
do not show ranges in hits: ☐

The above Results the shows the motif region present in the GBA protein, yellow colour indicates motif region and green colour indicates domain regions and the position start 143 ends with 162.

5.HOMOLOGUS MODELING: CPH MODELING



The above results show the CPH modeling server:

- Pink colour indicates helix.
- Yellow colour indicates sheets.
- Blue colour indicates turns.
- White colour indicates coil regin in glucosidase beta acid (GBA).

6. BIOPHYSICAL CHARACTERISATION:

DIPOLE MOVEMENT SERVER

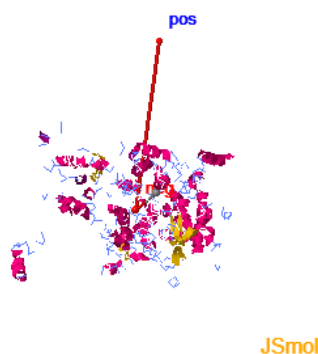
Dipole moment for

	No. of Chains=1		Spherical							
	No.Atoms	No.Res.	R _M	Pos.Res.	Neg.Res.	Charge	Dipole	Quadrupole	Crg./Nat.	Dip./Nat.
Value	9293.	1170.	1137.33	132.	145.	-12.	2804.	11272.	-0.0013	0.3018
No.Dev.Units	6.41	6.33	5.81	6.40	5.93	-1.14	5.41	2.64	0.02	-0.24

 Dipole vector (in atomic units): 61.54 434.93 -384.64

 Mass Moments vector: 2321.97 1797.00 1738.08

 Open a larger Jmol window.



The above results shows the dipole and mass moment vectors in, here red and grey-green line shows biophysical nature of proteins, respectively.

CONCLUSION

Gaucher disease is a rare, inherited disorder. It is a type of lipid metabolism disorder. If you have it, you do not have enough of an enzyme called glucocerebrosidase. This causes too much of a fatty substance to build up in your spleen, liver, lungs, bones and, sometimes, your brain. This prevents these organs from working properly. Glucocerebrosidase enzyme activity is stimulated by interaction with the lipid phosphatidylserine and the protein saposin C. Structural predictions (based on hydrophobic cluster analysis) indicate that the glutamine residues 235 and 340 play key roles in the active site of human glucocerebrosidase. Glucocerebrosidase is a lysosomal membrane-associated glycoprotein. Abnormal gene product. *GBA* pathogenic variants result in mRNA instability and/or loss of protein, or in an enzyme with altered activity and/or conformation. The protein sequence of glucosidase beta acid was retrieved from NCBI data base. The protein modeling and characterization of were analyzed using in silico bioinformatics tools. The present's studies revealed a number of interesting facts findings. Hence, we conclude that in future. The results should be used in drug designing process.

REFERENCES

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