

SUPPLEMENTAL DATA

Supplemental Figure S1

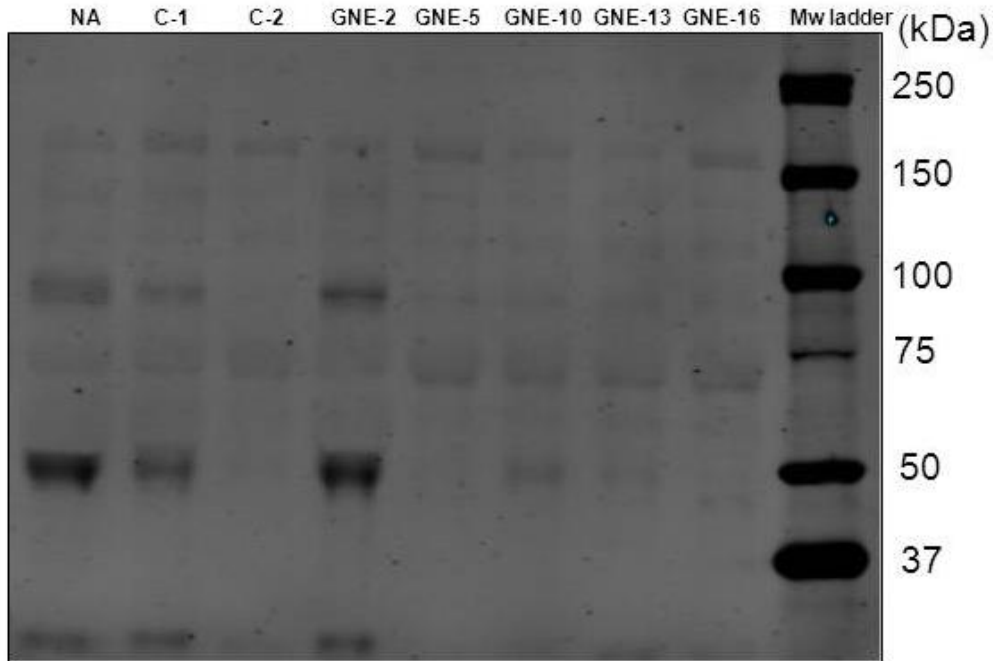
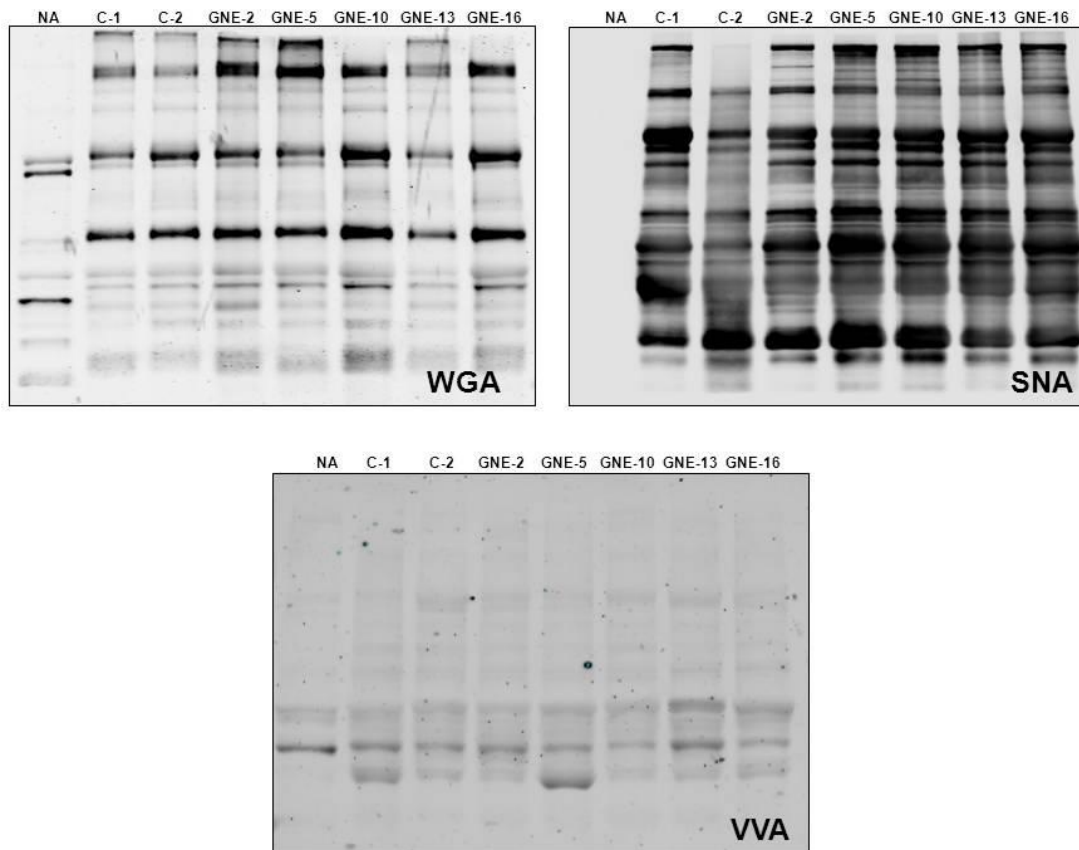


Figure S1: NCAM (RNL-1) immunoblotting of serum glycoproteins.

Serum samples (20 μ g) from neuraminidase treated control (NA), control (C-1, C-2), and GNE myopathy patients were immunoblotted with NCAM antibodies (RNL-1; sc-53007), as described by Valles-Ayoub et al. [25]. In contrast to the immunoblotting with the NCAM-H300 antibody (see **Figure 1**), immunoblotting with the NCAM-RNL-1 antibody does not show a different banding pattern in serum of GNE myopathy patients compared to control serum. Note that neuraminidase treated control serum did not show a different banding pattern either.

Supplemental Figure S2



Supplementary Figure S2. Western blotting followed by lectin staining of serum glycoproteins. Control (C-1, C-2), Neuraminidase treated control (NA), and GNE myopathy patients (GNE-2, -5,-10, -13,-16) serum was electrophoresed on SDS-PAGE gels, followed by electroblotting on nitrocellulose membranes. The membranes were incubated with the lectins SNA, WGA, or VVA. 10 μ g of total serum protein was loaded in the WGA and SNA labeled blots, and 20 μ g total serum protein was loaded in the VVA labeled blot. Note that the NA-treated control samples showed the expected reduction (for WGA and SNA) or increase (for VVA) in lectin binding, no significant differences in binding were present in GNE myopathy patients' compared to control samples.

Supplemental Table S1. Absolute T- and ST-values.

(normal ranges are highlighted in gray)

	T/ST	T-(uM)	ST-(uM)
	Normal Range		
	0.013-0.052	0.280-1.398	14.145-30.373
control1	0.03600597	0.88012959	24.44399043
control2	0.024737589	0.588552916	23.79184665
control3	0.050357143	0.761339093	15.1187905
control4	0.032507317	0.642548596	19.76627584
control5	0.025486765	0.411987041	16.16474456
control6	0.031530351	0.750539957	23.8037295
control7	0.029221739	0.772138229	26.42341804
control8	0.046083558	0.664146868	14.4117967
control9	0.049351003	1.398488121	28.33758262
control10	0.038265244	0.88012959	23.00075734
control11	0.030472441	0.696544276	22.85817135
control12	0.027367188	0.610151188	22.29499059
control13	0.046306667	0.745140389	16.09142792
control14	0.018971846	0.524298056	27.63558443
control15	0.037186567	1.069114471	28.75001788
control16	0.038818182	0.6587473	16.97007097
control17	0.024343887	0.530777538	21.80331903
control18	0.035924419	0.599352052	16.68369524
control19	0.049971264	0.761339093	15.23553791
control20	0.025650000	0.510259179	19.89314539
control21	0.021967647	0.415766739	18.92632095
control22	0.030728933	0.772138229	25.12740157
control23	0.024683866	0.471382289	19.09677734
control24	0.030382483	0.782937365	25.76936702
control25	0.025054667	0.534017279	21.31408435
control26	0.044735941	0.923326134	20.63947034
control27	0.025197952	0.57775378	22.92860049
control28	0.030213308	0.874730022	28.9518123
control29	0.035377778	1.074514039	30.37257019
control30	0.024160494	0.556155508	23.01921109
control31	0.017500000	0.279697624	15.98272138
control32	0.024002206	0.435205184	18.13188278
control33	0.019414154	0.387149028	19.94158649
control34	0.012814286	0.336933045	26.29354869

control35	0.024096774	0.63174946	26.21717973
control36	0.019197519	0.350431965	18.25402355
control37	0.030564741	0.858531317	28.08894459
control38	0.048591633	0.93412527	19.223994
control39	0.052225564	0.815334773	15.61179453
control40	0.032775000	0.534017279	16.29343337
control41	0.040855824	0.701943844	17.1809984
control42	0.035728827	0.63174946	17.68178546
control43	0.034503188	0.842332613	24.41318208
control44	0.032090347	0.723542117	22.54703345
control45	0.028532927	0.712742981	24.97966594
control46	0.037646341	0.680345572	18.07202363
Control47	0.029976062	0.534017279	17.81479103
Control48	0.047432705	0.799136069	16.84778604
Control49	0.045426647	0.642548596	14.14475077
Control50	0.043903104	0.93412527	21.27697553
mean	0.032966801	0.684082073	21.1724522
SD	0.009865643	0.211104208	4.441626615

Patient	GNE allele 1	GNE allele 2	T/ST	T (uM)	ST (uM)
				Normal Range	
			<0.052	0.280-1.398	14.145-30.373
GNE-2	c.1909+5G>A	p.V696M	0.10652626	1.34449244	12.62123039
GNE-5	p.M712T	p.M712T	0.07277276	1.02051836	14.02335691
GNE-10	p.D213V	p.V696M	0.10549956	1.24190065	11.77161965
GNE-13	p.M712T	p.M712T	0.13382721	1.08531317	8.10980918
GNE-14	p.V216A	p.A631V	0.06943573	0.84773218	12.20887551
GNE-16	p.M712T	p.M712T	0.09924569	1.65766739	16.70266379
GNE-20	p.W513X	p.A631V	0.07734388	1.18250540	15.28893296
GNE-21	p.D378Y	p.A631V	0.08537143	0.88552916	10.37266416
GNE-28	p.R129X	p.V696M	0.09140743	1.56317495	17.10118034
GNE-32	p.M712T	p.M712T	0.06578169	0.98002160	14.89809162
GNE-980	p.M712T	p.M712T	0.10246480	1.36609071	13.33229279
GNE-981	p.M712T	p.M712T	0.11202826	1.53347732	13.68830792
GNE-914a	p.M712T	p.M712T	0.10040766	0.22084233	2.19945707
GNE-914b*	p.M712T	p.M712T	0.04536585	0.40172786	8.85529158