Bloom Syndrome - **Gene:** BLM. **Variants (1):** 2281del6ins7.

Canavan Disease - **Gene:** ASPA. **Variants (4):** E285A, Y231X, A305E, IVS2-2A>G.

Cystic Fibrosis - **Gene:** CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, E92X, Y122X, G178R, R347H, Q493X, V520F, S549N, P574H, M1101K, D1152H, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K.

Familial Dysautonomia - **Gene:** IKBKAP. **Variants (2):** IVS20+6T>C, R696P.

Fanconi Anemia Type C - **Gene:** FANCC. **Variants (3):** IVS4+4A>T, 322delG, R548X.

Gaucher Disease - **Gene:** GBA. **Variants (10):** N370S, L444P, 84GG, IVS2+1G>A, V394L, R496H, D409H, D409V, R463C, R463H.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. **Variants (28):** Hb S, K17X, Q39X, Phe41fs, Ser9fs, IVSII-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. **Variants (9):** 1278insTATC, IVS12+1G>C, G269S, IVS9+1G>A, R178H, IVS7+1G>A, 7.6kb del, G250D, R170W.

Mucolipidosis IV - **Gene:** MCOLN1. **Variants (2):** 511_6944del, IVS3-2A>G.

Niemann-Pick Disease, SMPD1-Associated - **Gene:** SMPD1. **Variants (4):** fsP330, L302P, R496L, p.R608del.

Spinal Muscular Atrophy - **Gene:** SMN1. **Variants (1):** SMN1 copy number.

Beta Thalassemia - **Gene:** HBB. **Variants (27):** K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab.