

Samples Available from NIH:

Diagnosis	Cell line	Plasma	Serum	CSF	Urine
Late Onset Tay-Sachs	√	√	√	√	√
Infantile Tay-Sachs	√				
Juvenile Tay-Sachs		√	√	√	√
Infantile Sandhoff					
Juvenile Sandhoff	√	√	√		√
Late Onset Sandhoff	√	√	√	√	√
Infantile GM1					
Juvenile GM1					
Late Onset GM1					
Canavan					

Genetic Variants:

HEXA

- c.805G>A (p.Gly269Ser)
- c.1274_1277 dup (p.Y427Ifs*5)
- c.806-7G>A
- c.1A>T
- c.986+5G>A
- c.533G>A (p.Arg178His)
- c.1073+1G>A
- c.1421+1G>C
- c.929_930delCT (p.Ser310fs)

HEXB

- c.1514G>A
- c.1417+5G>A
- c.1417G>A (p.Gly473Ser)