

Biomarker for Niemann-Pick Type C1 Disease by LC-MS/MS

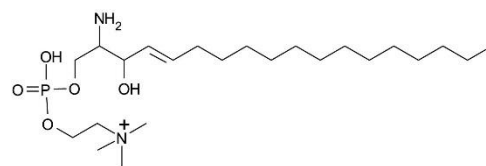
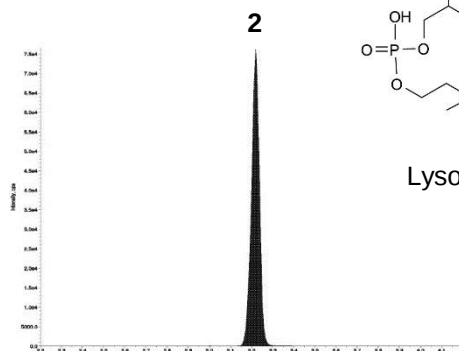
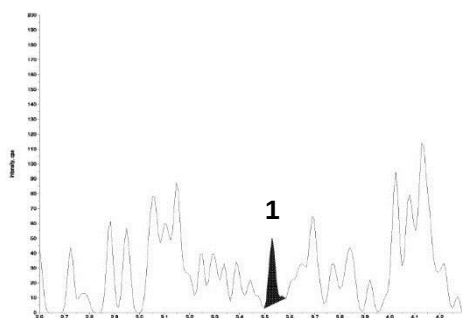
Conditions

Column: ACE 3 C8
Dimensions: 50 x 2.1 mm
Part Number: ACE-112-0502
Mobile Phase: A: 50 mM formic acid in H₂O
B: 50 mM formic acid in MeCN/acetone (1:1 v/v)

Time (mins)	%B
0.0	5
4.0	66
4.1	100
5.1	100
5.9	5

Flow Rate: 0.9 mL/min
Injection: 5 µL
Temperature: 60 °C
Detection: API 4000 triple quad MS
ESI in positive ion mode
Temperature: 500 °C

Human Control Plasma

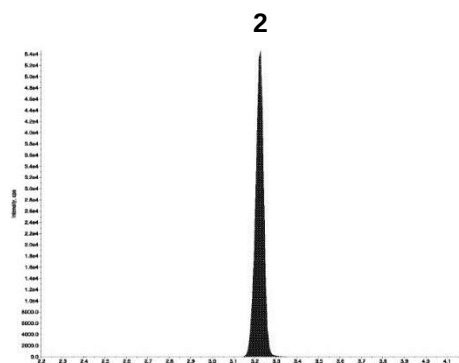
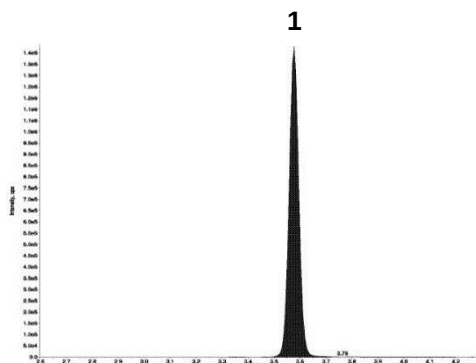


Lyso-SM (lyso-sphingomyelin)

1. Lyso-SM-509
(*m/z* 509 → 184)

2. Lyso-Gb2 (IS)
(*m/z* 624 → 282)

Niemann-Pick Patient Sample



Giese A, Mascher H, Grittner U, Eichler S, Kramp G, Lukas J, te Vrugte D, Eisa N, Cortina-Borja M, Porter F, Platt F, Rolfs A. Orphanet Journal of Rare Diseases (2015) 10:78 A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. DOI 10.1186/s13023-015-0274-1

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