

# Lyso GB1 (glycosylsphingosine) in plasma and dry blood spots in lysosomal storage diseases

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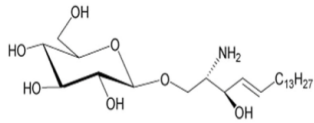
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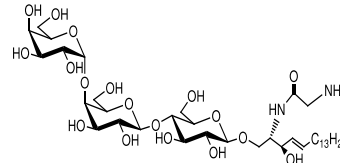
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# “Lysos” are good diagnostic markers of LSD

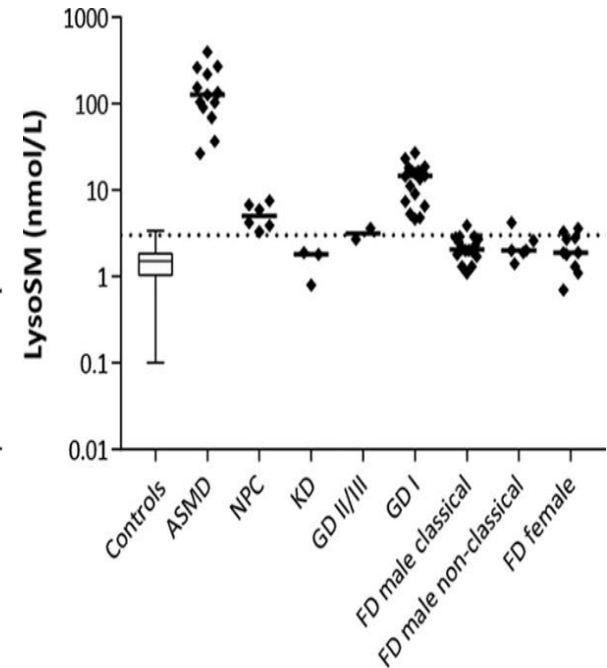
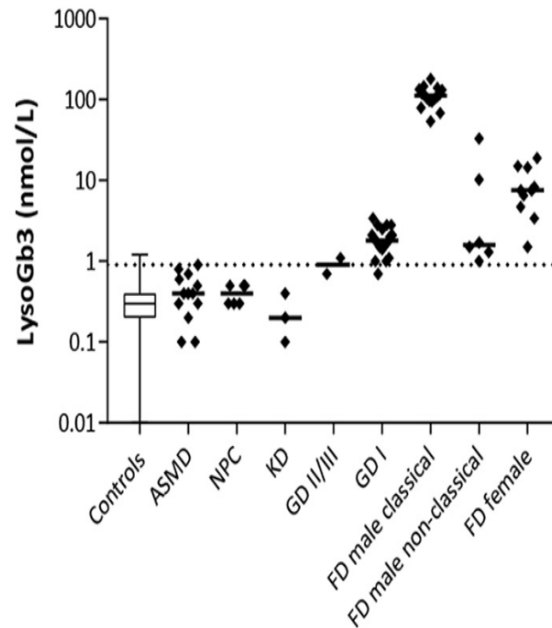
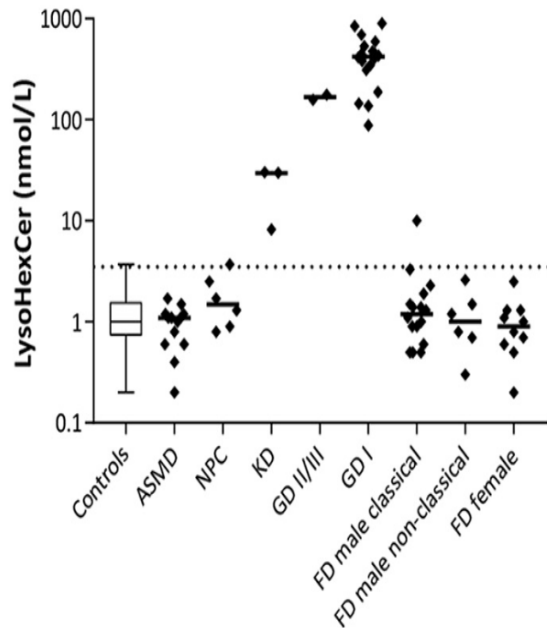
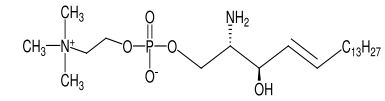
Lyso-Gb1 / GalSph (KD, GD)



Lyso-Gb3 (Fabry)



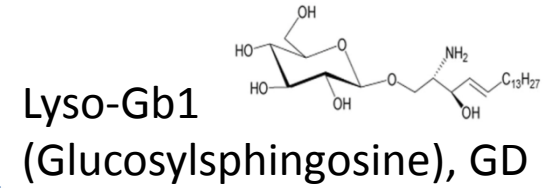
Lyso-SM (NP-A, NP-B)



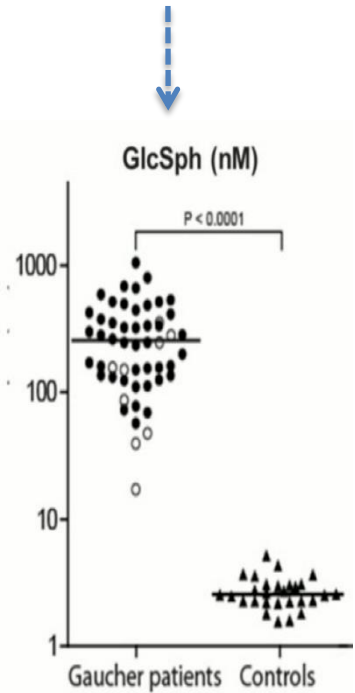
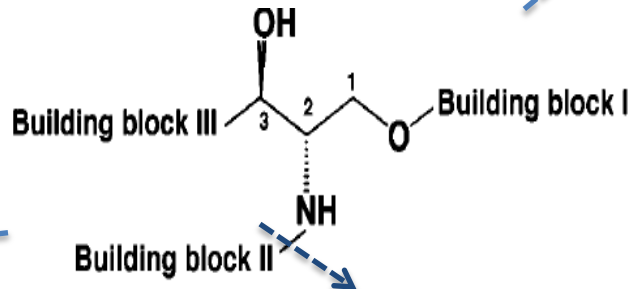
# Breaking Bad or how to make soluble what is insoluble ?

Lysosomal enzymes

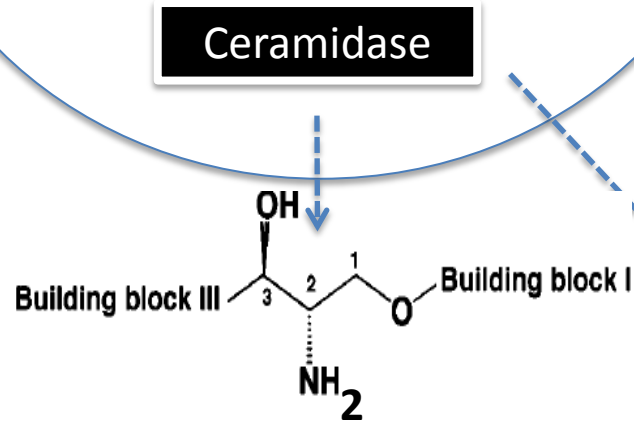
Lyso-SM (NPA, NPB)



Lyso-Gb3  
(Fabry)

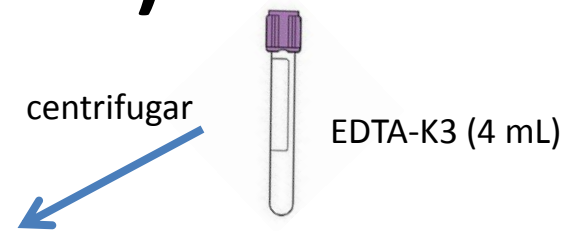
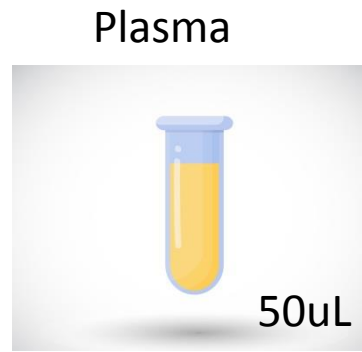
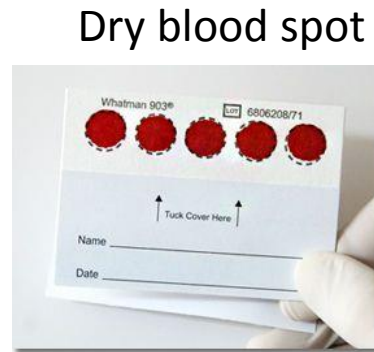
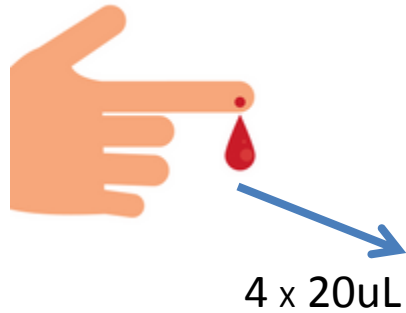


Lyso-Sulf (MLD) ?



Galactosylsphingosine (Krabbe's)

# Methodology (LC-MS/MS)

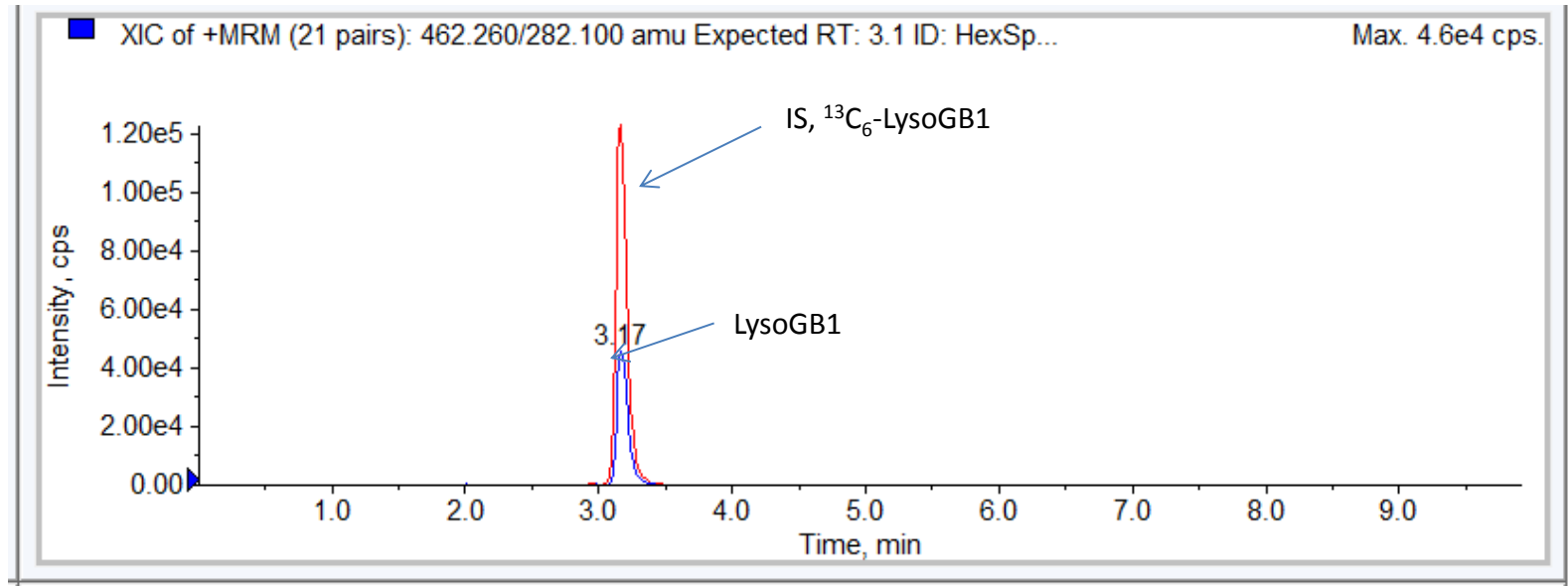


Internal stadard,  $^{13}\text{C}$ -LysoGB1

Lipid extraction

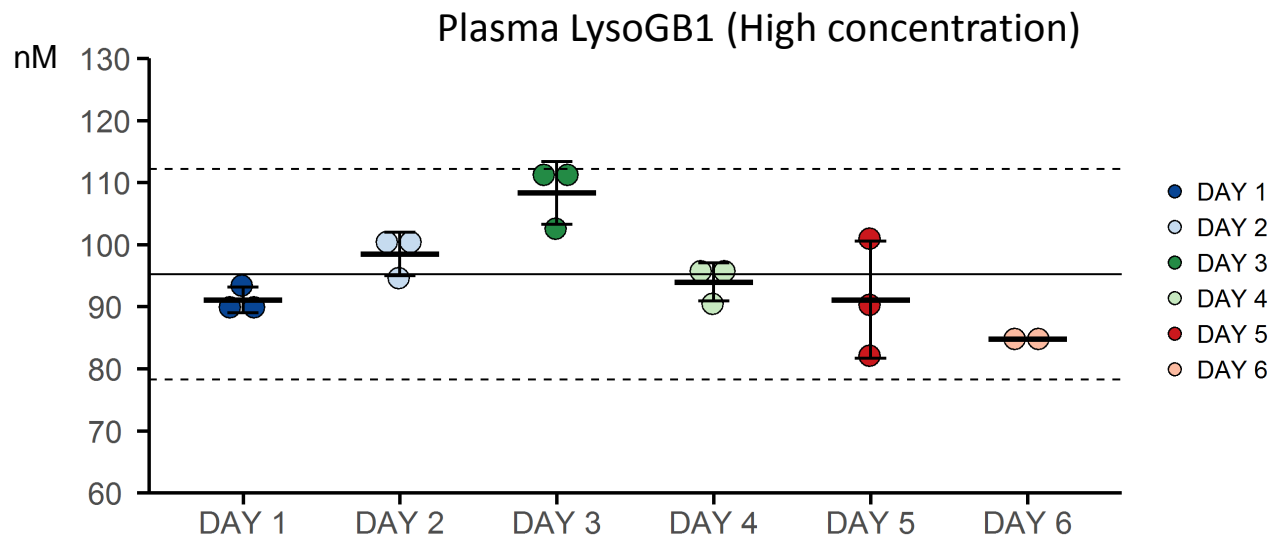
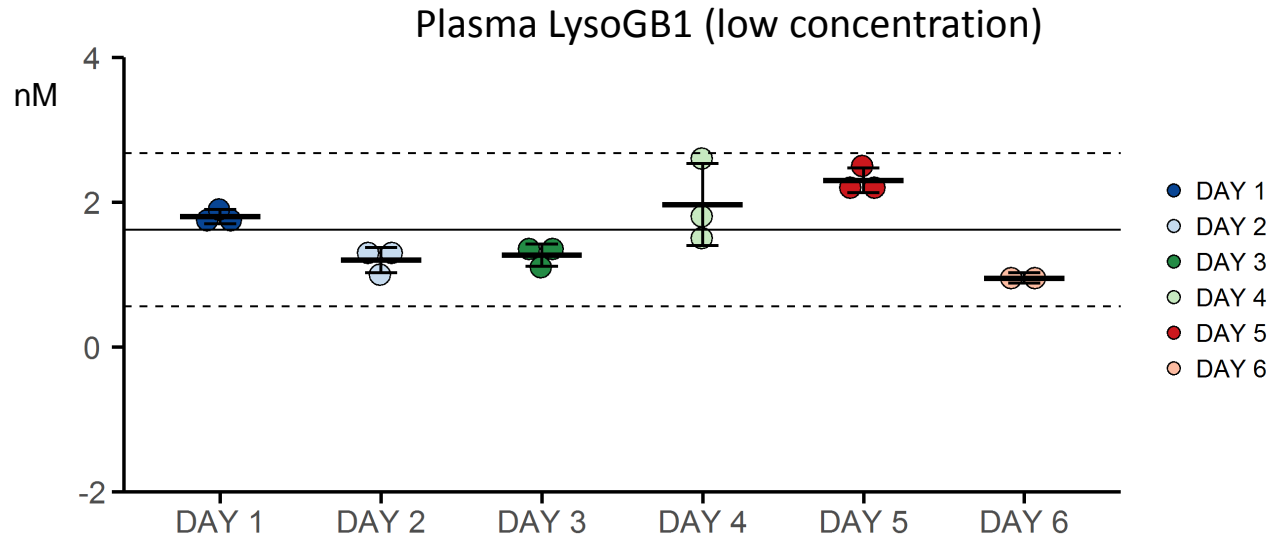


# Methodology (LC-MS/MS)

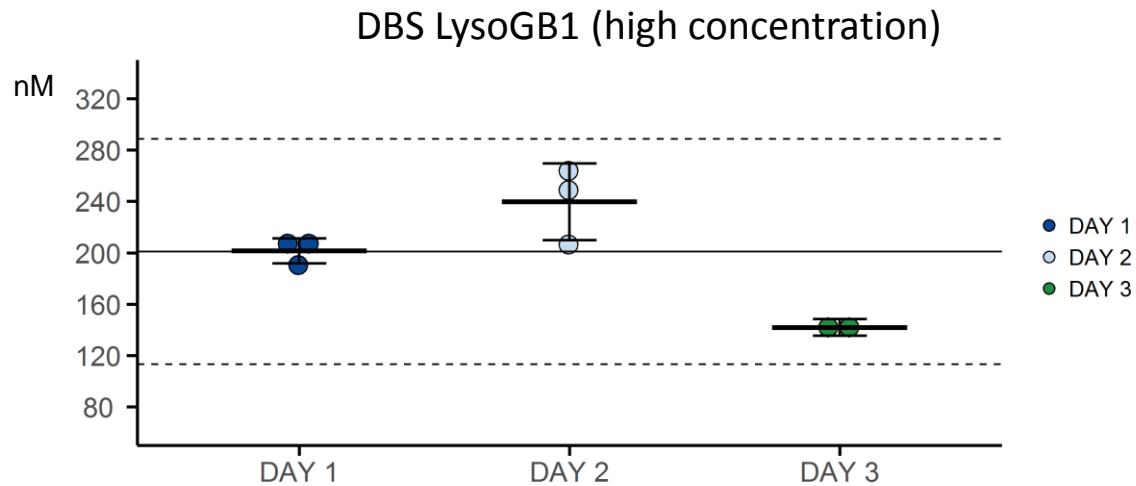
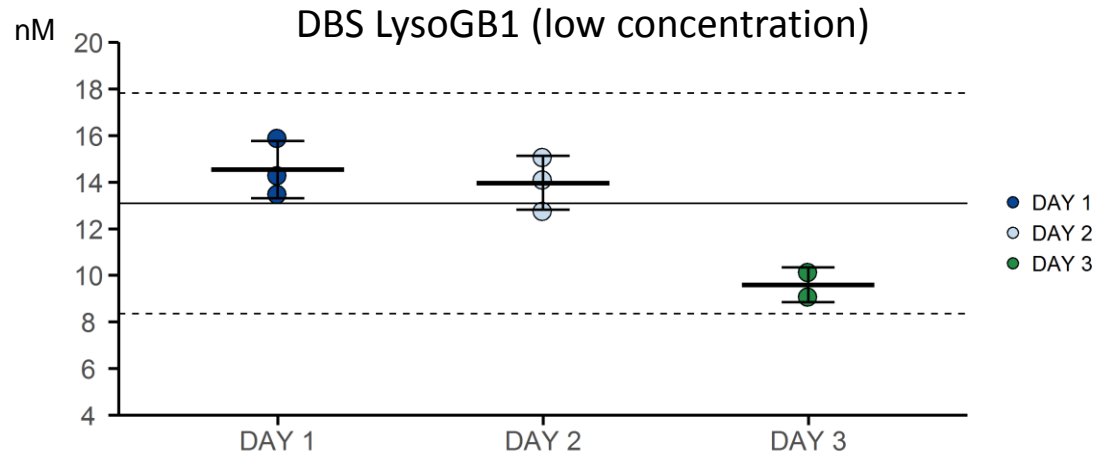


	Q1 Mass (Da)	Q3 Mass (Da)	Time (min)	ID	DP (volts)	CE (volts)	CXP (volts)	
Quant →	9	462.260	282.100	3.09	HexSph	76.000	29.000	16.000
	10	462.260	264.100	3.09	HexSph_2	76.000	25.000	14.000
IS →	11	468.500	282.100	3.09	IS_13CGluSph	101.000	29.000	16.000
	12	468.500	264.200	3.09	13CGluSph_3	96.000	25.000	14.000

# Quality control in plasma samples



# Quality control in plasma samples in dry blood spots (DBS)



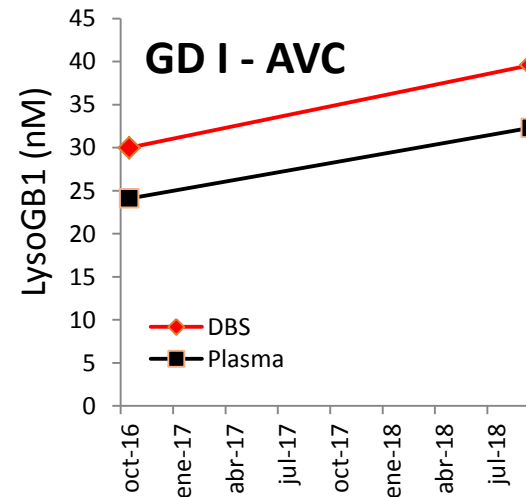
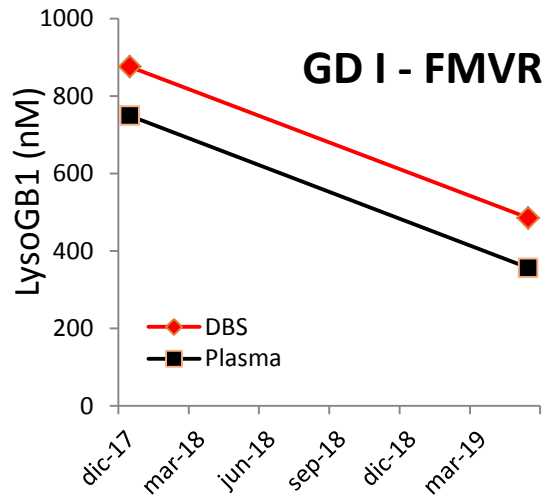
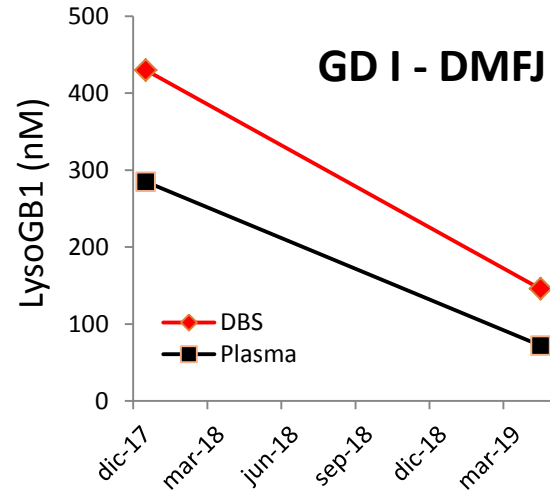
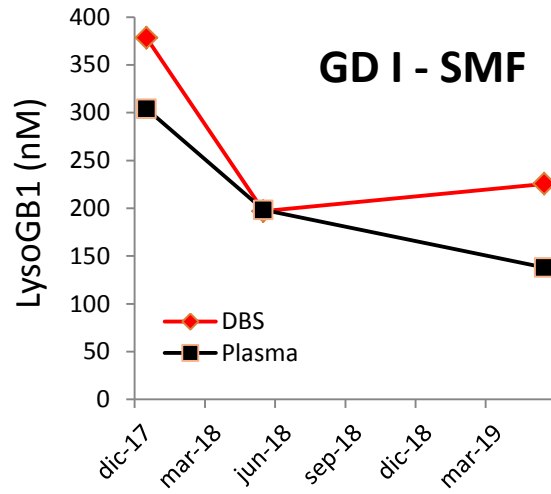
# LysoGB1: our experience

	Glucosylsphingosine		CT Activity (nmol/mL.h)	CCL18 (ng/mL)
	Plasma (nM)	Dry blood (nM)		
	$\leq 12$	$\leq 47$	(4-76)	(18.9-106.3)
GD type I	<b>284.9</b> ± 8.5	<b>430.15</b> ± 45.0	5116	829.2
GD type I	<b>304.1</b> ± 19.9	<b>378.35</b> ± 10.4	2845.1	421.5
GD type I (treated)	<b>22.3</b> ± 0.4			
GD type I (treated)	<b>24.1</b> ± 1.4			
GD type I	<b>748.8</b> ± 46.4	<b>875.0</b> ± 24.4	9391.2	1264
GD type I	<b>256.8</b> ± 33.9			
GD type III		<b>50.9</b> ± 3.4		
Familiar (GD I)	1.8 ± 0.1			
Familiar (GD I)	2.3 ± 0.2			
Familiar (GD I)	2.2 ± 0.0			
Familiar (GD I)	1.2 ± 0.0			

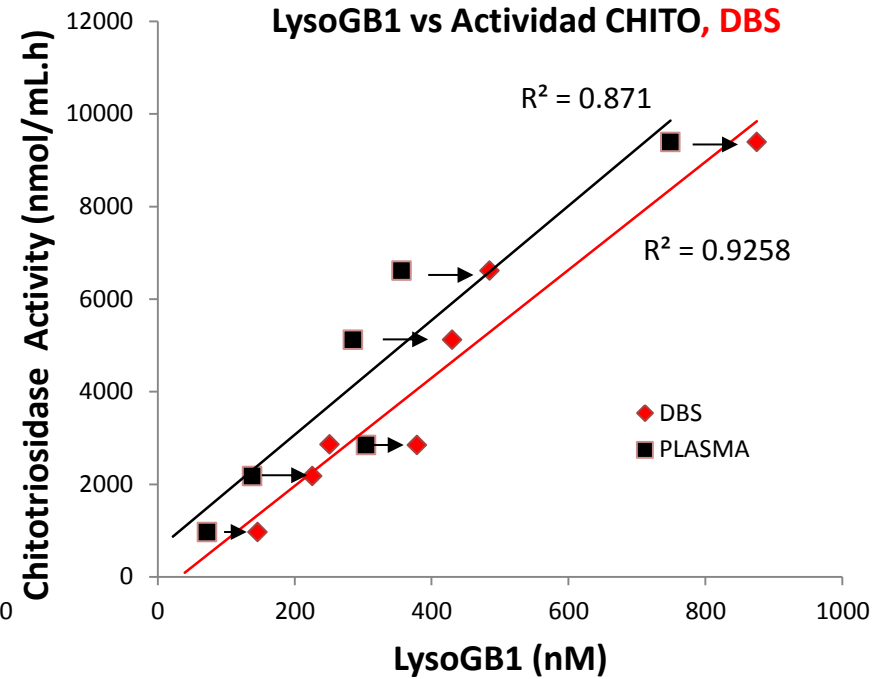
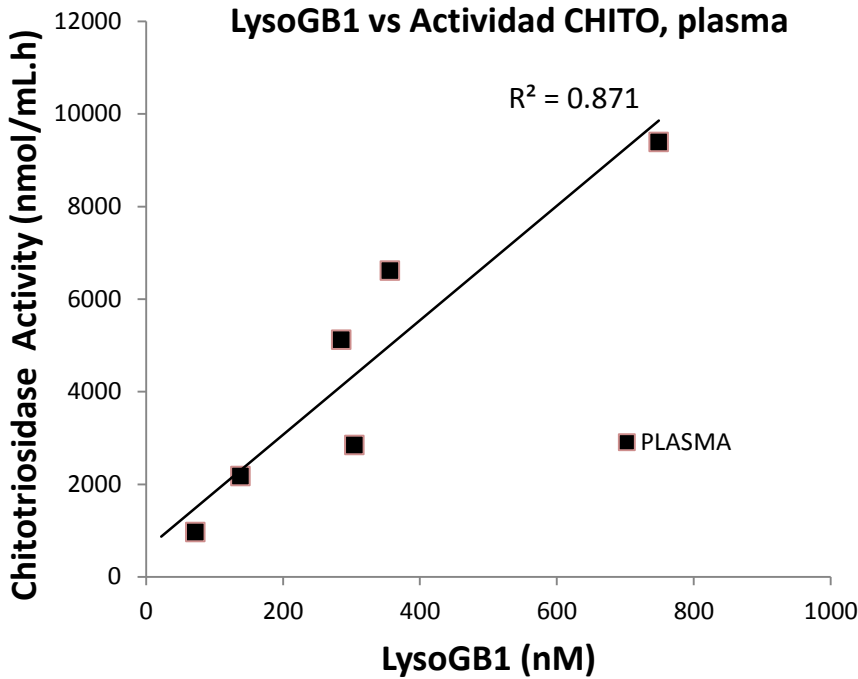
LysoGB1      **Sensitivity 100 %**  
**Especificity 100 %**



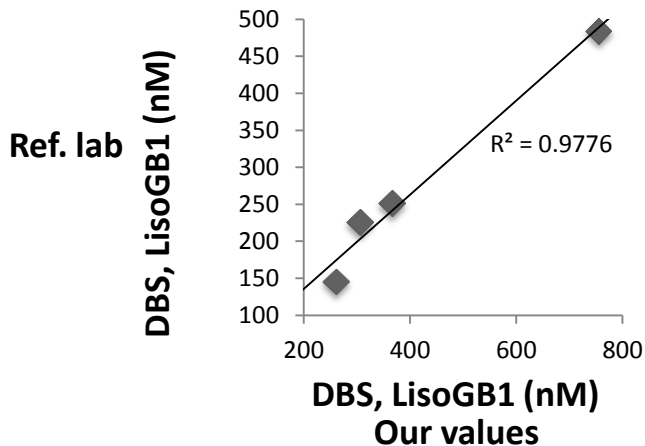
# LysoGB1: our experience II



# LysoGB1: correlation



## Correlación con Laboratorio de Referencia



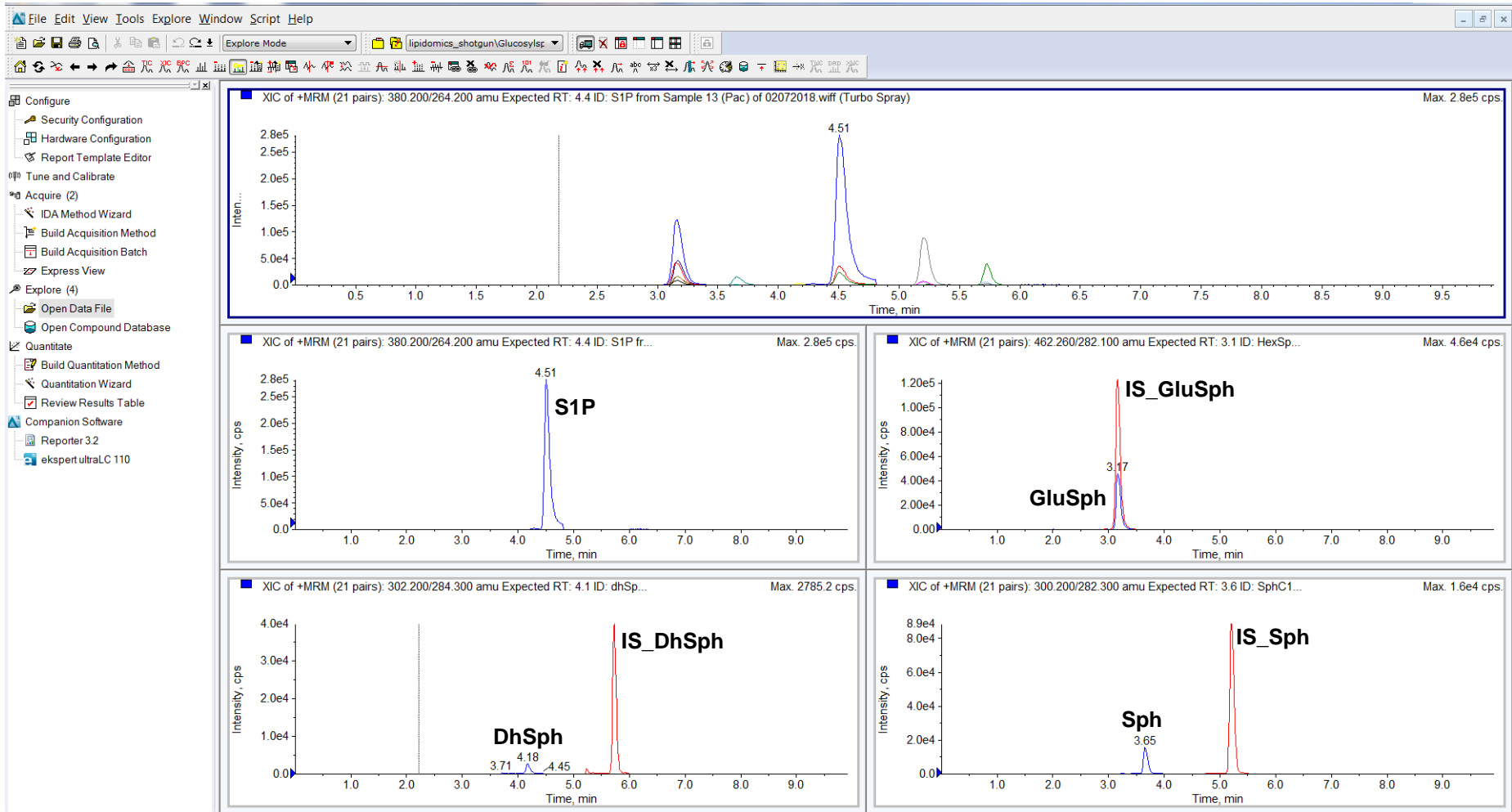
$$\text{LysoGB1}_{(\text{DBS})} - \text{LysoGB1}_{(\text{Plasma})} \approx 80 \text{ nM}$$

ng/mL

Dividir 0.46  $\uparrow$   $\downarrow$  Multiplicar 0.46

nmol/L (nanoMolar)

# Working on other metabolites



# Contact information

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