

Source	Method	Assay	Results	BGAL
Blood	DBS*	AGLU-4MUG+8ACB	<b>85.3</b>	4710
		Normal range (pmol/punch/17 hrs) Patient range ( pmol/punch/17hrs)	94 - 448 11 - 56	476 - 4680 88
	Leukocytes	AGLU-GN+3ACB	<b>- 0.7</b>	
		Normal range (nmol/hr/mg) Patient range ( nmol/ hr/mg)	>40 -250 0 - 3.5	
	DBS**	AGLU-4MUG+8ACB	<b>1.9</b>	
		Normal range (nmol/hr/mg) Patient range ( nmol/ hr/mg)	>6.7 - 27 1.1 - 4.9	
DBS**	AGLU-4MUG+8ACB	<b>68</b>	5230	
	Normal range (pmol/punch/17 hrs) Patient range ( pmol/punch/17hrs)	94 - 448 11 - 56	476 - 4680 88	
Skin biopsy	Fibroblast	AGLU-4MUG	<b>0.42</b>	
		Normal range (nmol/hr/mg) Patient range ( nmol/ hr/mg)	>45- 180 0 - 3	
Urine	TGLC:	Normal range (mmol/mol creatinine)	<b>60</b>	
		Age in years		
		< 1		2.4 - 16.2
		1-5		0 - 10.7
		6-10		0 - 3.6
		11-20		0 - 1.9
		>20		0 - 2.2
		Patient range (mmol/mol creatinine)		
		< 1		16.6 - 74
		1-5		27 - 66.7
		6-10		6 - 111
		11-20		3.4 - 160
		>20		2.3 - 130

TGLC concentrations are in millimoles per mol creatinine

**Patient 32**  
**Genotype:**  
 p.(Glu176Argfs\*45)<sup>1</sup>/<sub>2</sub>  
 p.(Gly828\_Asn882del)<sup>2</sup>  
**Clinical diagnosis:**  
 Classic Infantile Pompe disease