

## UNIVERSITY OF ALABAMA AT BIRMINGHAM HEALTH SERVICES FOUNDATION, PC

## Department of Genetics

## Lysosomal Storage Disease Services

634 Kaul Building, 720 20<sup>th</sup> Street South, Birmingham, AL 35233

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Available Tests	Disease	Source	Price	CPT Code #
1 Acetyl CoA: $\alpha$ -glucosaminide N-acetyltransferase	Sanfilippo syndrome, type C.....	FIB	\$550	82658/82657
2 Arylsulfatase A (plasma screen)	Mucopolidosis II or III.....	PLAS	\$200	82657
3 Arylsulfatase B	Maroteaux - Lamy.....	FIB	\$300	84311
4 $\alpha$ -Fucosidase	Fucosidosis.....	FIB, WBC, PLAS	\$300	84311
5 $\alpha$ -Galactosidase A	Fabry disease.....	FIB, WBC	\$500	84311
6 $\beta$ -Galactosidase	GM <sub>1</sub> -gangliosidosis / Morquio B.....	FIB, WBC	\$300	84311
7 $\alpha$ -Glucosidase	Pompe disease.....	FIB	\$500	82963
8 $\beta$ -Glucosidase	Gaucher disease.....	FIB	\$300	84311
9 $\beta$ -Glucuronidase	Sly syndrome.....	FIB, WBC	\$300	84311
10 Heparin sulfamidase	Sanfilippo syndrome, type A.....	FIB	\$550	82658/84311
11 $\beta$ -Hexosaminidase (total activity)	Sandhoff disease.....	FIB, WBC, PLAS	\$300	83080
12 $\beta$ -Hexosaminidase A	Tay-Sachs disease.....	FIB, WBC, PLAS	\$300	83080
13 Iduronate 2-sulfatase	Hunter syndrome.....	FIB, PLAS	\$550	82658/82657
14 $\alpha$ -Iduronidase	Hurler syndrome.....	FIB, WBC	\$300	84311
15 $\alpha$ -Mannosidase	Alpha-mannosidosis.....	FIB, WBC	\$300	84311
16 $\beta$ -Mannosidase	Beta-mannosidosis.....	FIB, WBC, PLAS	\$300	84311
17 N-acetylgalactosamine 6-sulfatase	Morquio syndrome, type A.....	FIB	\$550	82657
18 N-acetylglucosamine 6-sulfatase	Sanfilippo syndrome, type D.....	FIB	\$550	82657
19 N-acetyl- $\alpha$ -D-glucosaminidase	Sanfilippo syndrome, type B.....	FIB, PLAS	\$400	82657
20 Sialidase (neuraminidase)	Sialidosis.....	FIB	\$400	82657
21 N-acetyl- $\alpha$ -D-galactosaminidase	Schindler disease.....	FIB, WBC, PLAS	\$300	82657
22 Aspartylglucosaminidase	Aspartylglucosaminuria.....	WBC, FIB	\$400	82657
23 Quantitation of urinary mucopolysaccharides (This provides an extensive analysis of gags and includes..... glycosaminoglycans – measured by uronic acid and specific assay for heparan sulfate..... and electrophoresis which provides separation of each of the gag components).....		URINE	\$500	83864 82486/82485/82491 82664
24 Sialic Acid.....		URINE	\$400	82657
25 TLC (Oligosaccharides).....		URINE	\$300	84375
26 Sulfated Monosaccharide assay.....		URINE	\$300	82486
27 Develop a cell line from a skin biopsy or other tissue.....		-----	\$500	88233
28 Cryopreservation, freezing & storage of cells, each cell line (for 1 year only).....		-----	\$300	88240
29 Thawing and expansion of frozen cells, each vial of cells.....		-----	\$300	88241

FIB – cultured skin fibroblasts; White blood cells (WBC) and plasma (PLAS) are obtained from Sodium Heparinized Blood