

UNIVERSITY OF ALABAMA AT BIRMINGHAM HEALTH SERVICES FOUNDATION, PC

Department of Genetics

Lysosomal Storage Disease Services

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Available Tests	Disease	Source	Price	CPT Code #
1 Acetyl CoA: α -glucosaminide N-acetyltransferase	Sanfilippo syndrome, type C.....	FIB	\$550	82657/82658/84155
2 Arylsulfatase A (serum screen)	Mucopolidosis II or III.....	PLAS	\$200	82657/84311
3 Arylsulfatase B	Maroteaux - Lamy.....	FIB	\$300	82657/84311/84155
4 α -Fucosidase	Fucosidosis.....	FIB, WBC, PLAS	\$300	82657/84311/84155*
5 α -Galactosidase A	Fabry disease.....	FIB, WBC	\$500	82657/84311/84155
6 β -Galactosidase	GM ₁ -gangliosidosis / Morquio B.....	FIB, WBC	\$300	82657/84311/84155
7 α -Glucosidase	Pompe disease.....	FIB	\$500	82657/84311/84155
8 β -Glucosidase	Gaucher disease.....	FIB	\$300	82963/84155
9 β -Glucuronidase	Sly syndrome.....	FIB, WBC	\$300	82657/84311/84155
10 Heparin sulfamidase	Sanfilippo syndrome, type A.....	FIB	\$550	82657/82658/84155
11 β -Hexosaminidase (total activity)	Sandhoff disease.....	FIB, WBC, PLAS	\$300	83080/84155*
12 β -Hexosaminidase A	Tay-Sachs disease.....	FIB, WBC, PLAS	\$300	83080/84155*
13 Iduronate 2-sulfatase	Hunter syndrome.....	FIB, PLAS	\$550	82657/82658/84155*
14 α -Iduronidase	Hurler syndrome.....	FIB, WBC	\$300	82657/84311/84155
15 α -Mannosidase	Alpha-mannosidosis.....	FIB, WBC	\$300	82657/84311/84155
16 β -Mannosidase	Beta-mannosidosis.....	FIB, WBC, PLAS	\$300	82657/84311/84155*
17 N-acetylgalactosamine 6-sulfatase	Morquio syndrome, type A.....	FIB	\$550	82657/84311/84155
18 N-acetylglucosamine 6-sulfatase	Sanfilippo syndrome, type D.....	FIB	\$550	82657/84311/82542 84155
19 N-acetyl- α -D-glucosaminidase	Sanfilippo syndrome, type B.....	FIB, PLAS	\$400	82657/84311/84155 *
20 Sialidase (neuraminidase)	Sialidosis.....	FIB	\$400	82657/84311/84155
21 N-acetyl- α -D-galactosaminidase	Schindler disease.....	FIB, WBC, PLAS	\$300	82657/84311/84155*
22 Aspartylglucosaminidase	Aspartylglucosaminuria.....	WBC, FIB	\$400	82657/84311/84155
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(x3) 84311(x3)/82664
24 Sialic Acid.....		URINE	\$400	84275/84311
25 TLC (Oligosaccharides).....		URINE	\$300	84375
26 Sulfated Monosaccharide assay.....		URINE	\$300	82542/84311
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

* Use 84155 only with Fibroblasts or WBC*