



The University of Chicago Genetic Services Laboratories

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 CLIA #: 14D0917593 CAP #18827-49

QUICK GUIDE TO GENETIC TESTING

TEST	DISORDER	CPT CODES	TAT	COST
CYTOGENETIC and FISH tests: 3 to 10cc blood in a sodium heparin/green top tube				
Chromosome analysis - amniotic fluid		88235, 88269, 88291	Prelim: 5 days, Final: 10 days	\$900
AneuVysion™ - amniotic fluid		88271, 88275, 88291	24 to 48 hours	\$375
Chromosome analysis - chorionic villi		88235, 88267, 88291	Prelim: 5 days, Final: 10 days	\$1,000
Chromosome analysis - blood		88230, 88262, 88291	12 days, STAT analysis available	\$700
Chromosome analysis - tissue		88233, 88262, 88291	12 days	\$750
MICRODELETION OR MICRODUPLICATION TESTING for Prader-Willi/Angelman, SNRPN duplication [autism], proximal 15q polymorphism, lissencephaly, DiGeorge, Williams, Sotos, CHARGE, SRY, cri-du-chat, Wolf-Hirschhorn, Xist				
3 to 10cc blood in a sodium heparin/green top tube				
FISH - Culture and first probe		88230, 88271, 88291, 88273	10 - 12 days	\$325
FISH - additional probe		88271	10 - 12 days	\$150
TelAll (subtelomeric screening)		88230, 88271 x 3, 88273, 88291	14 - 21 days	\$950
Angelman syndrome and Prader-Willi syndrome testing: 3 to 10cc of blood in an EDTA/purple top tube				
M-PCR		83891, 83898, 83894, 83912	2 - 4 weeks	\$315
FISH - 15q11**requires sodium heparin/green top tube		88230, 88271, 88291, 88273	10 - 12 days	\$325
UPD 15 testing		83891, 83898 x 4, 83894, 83912	2 - 4 weeks	\$540
Imprinting Center (IC) deletion analysis		83891, 83900, 83901, 83912	2 - 4 weeks	\$450
UBE3A sequencing	Angelman syndrome	83891, 83898 x 4, 83904 x 9, 83912	4 - 6 weeks	\$2,025
Brain Malformation testing: 3 to 10cc of blood in an EDTA/purple top tube				
Lissencephaly panel--TIER 1 (LIS1 & DCX sequencing and deletion/duplication testing)		83891, 83898 x6, 83904 x11, 83900, 83901, 83912	6 - 8 weeks	\$2,825
Lissencephaly panel--TIER 2 (ARX sequencing)		83891, 83898 x2, 83904 x4, 83912	4 - 6 weeks	\$1,000
ARX sequencing	XLAG	83891, 83898 x2, 83904 x4, 83912	4 - 6 weeks	\$1,000
LIS1 mutation analysis (sequencing and deletion/duplication testing)	lissencephaly	83891, 83898 x3, 83904 x7, 83900, 83901 x2, 83912	4 - 6 weeks	\$2,000
LIS1 sequencing	lissencephaly	83891, 83898 x4, 83904 x7, 83912	4 - 6 weeks	\$1,675
DCX mutation analysis (sequencing and deletion/duplication testing)	X-linked lissencephaly	83891, 83898 x2, 83904 x5, 83900, 83901 x2, 83912	4 - 6 weeks	\$1,575
DCX(XLIS) sequencing	X-linked lissencephaly	83891, 83898 x3, 83904 x5, 83912	4 - 6 weeks	\$1,250
LIS1/DCX deletion/duplication analysis	X-linked lissencephaly	83891, 83900, 83912	4 weeks	\$350
GPR56 sequencing	bilateral frontoparietal polymicrogyria	83891, 83898 x 4, 83904 x 6, 83912	4 - 6 weeks	\$1,500
FISH - 17p13**requires sodium heparin/green top tube	lissencephaly	88230, 88271, 88291, 88273	10 - 12 days	\$325
Infantile spasms/atypical Rett syndrome testing: 3 to 10cc of blood in an EDTA/purple top tube				
Infantile spasms/atypical Rett syndrome panel (MECP2 sequencing and deletion analysis, ARX & CDKL5 sequencing)		83891, 83898 x 8, 83904 x 16, 83900, 83901 x 2, 83912	6 - 8 weeks	\$3,950
MECP2 sequencing		83898 x 3, 83904 x 4	4 - 6 weeks	\$925
MECP2 deletion analysis		83891, 83900, 83901 x 2, 83912	4 weeks	\$550
ARX sequencing		83891, 83898 x2, 83904 x4, 83912	4 - 6 weeks	\$1,000
CDKL5/STK9 sequencing		83891, 83898 x 4, 83904 x 9, 83912	4 - 6 weeks	\$2,025
MECP2 testing: 3 to 10cc of blood in an EDTA/purple top tube				
MECP2 mutation analysis (sequencing and del/dup testing)	Rett syndrome	83891, 83898 x2, 83904 x4, 83900, 83901 x2, 83912	4 - 6 weeks	\$1,400
MECP2 sequencing exons 1-4	Rett syndrome	83898 x 3, 83904 x 4	4 - 6 weeks	\$925
* MECP2 sequencing exon 1 only	Rett syndrome	83904	2 - 3 weeks	\$175
MECP2 deletion/duplication analysis for females	Rett syndrome	83891, 83900, 83901 x 2, 83912	4 weeks	\$550
MECP2 duplication analysis--males	XL mental retardation	83891, 83898 x 4, 83912	4 weeks	\$450



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Chondrodysplasia punctata testing: 3 to 10cc of blood in an EDTA/purple top tube				
ARSE sequencing	X-linked recessive (CDPX1)	83891, 83898 x 4, 83904 x 6, 83912	4 - 6 weeks	\$1,500
EBP sequencing	X-linked dominant (CDPX2)	83891, 83898 x5, 83904 x2, 83912	4 - 6 weeks	\$875
Cornelia de Lange syndrome testing: 3 to 10cc of blood in an EDTA/purple top tube				
NIPBL sequencing	Cornelia de Lange syndrome	83891, 83898 x9, 83904 x9, 83912	10 weeks	\$2,400
NIPBL deletion/duplication analysis		83891, 83900, 83912	4 weeks	\$350
SMC1A sequencing		83891, 83898 x 4, 83904 x 9, 83912	4 - 6 weeks	\$2,025
MCT8 testing: 3 to 10cc of blood in a red top tube and 3 to 10cc of blood in a purple top (EDTA) tube				
Normal thyroid testing only	Allan-Herndon-Dudley syndrome/MCT8-related thyroid hormone cell transporter deficiency	83891, 83912, 90001	2 weeks	\$350
Thyroid testing and MCT8 sequencing		83891, 83898 x 4, 83904 x 6, 83912	4 - 6 weeks	\$1,500
Menkes disease testing: 3 to 10cc of blood in an EDTA/purple top tube				
ATP7A mutation analysis (sequencing and del/dup testing)	Menkes disease	83891, 83898 x3, 83904 x9, 83900, 83901 x2, 83912	4 - 6 weeks	\$2,350
ATP7A sequencing		83891, 83898 x4, 83904 x9, 83912	4 - 6 weeks	\$2,025
ATP7A deletion/duplication analysis		83891, 83900, 83912	4 weeks	\$350
Neurodegeneration with brain iron accumulation (NBIA) testing: 3 to 10cc of blood in an EDTA/purple top tube				
PANK2 sequencing	PKAN/Hallervorden-Spatz syndrome	83891, 83898 x4, 83904 x5, 83912	4 - 6 weeks	\$1,325
PLA2G6 sequencing	infantile neuroaxonal dystrophy	83891, 83898x4, 83904x9, 83912	4 - 6 weeks	\$2,025
Rubinstein-Taybi syndrome testing: 3 to 10cc of blood in an EDTA/purple top tube				
CREBBP sequencing	Rubinstein-Taybi syndrome	83891, 83898 x9, 83904 x9, 83912	10 weeks	\$2,400
CREBBP deletion/duplication analysis		83891, 83900, 83912	4 weeks	\$350
Sotos syndrome testing: 3 to 10cc of blood in an EDTA/purple top tube				
NSD1 sequencing	Sotos syndrome	83891, 83898 x9, 83904 x9, 83912	10 weeks	\$2,400
NSD1 deletion/duplication analysis		83891, 83900, 83912	4 weeks	\$350
FISH - 5q35**requires sodium heparin/green top tube		88230, 88271, 88291, 88273	10 - 12 days	\$325
UGT1A1 testing: 3 to 10cc of blood in an EDTA/purple top tube				
UGT1A1 genotyping for Gilbert syndrome		83891, 83898, 83894, 83912	2 weeks	\$383
UGT1A1 genotyping for irinotecan dosing		83891, 83898, 83912	1 week	\$293
Other DNA tests: 3 to 10cc of blood in an EDTA/purple top tube				
UPD testing - chromosomes 6, 7, 14 or 15	transient neonatal diabetes, Russell-Silver syndrome, UPD14, Angelman syndrome or Prader-Willi syndrome	83891, 83898 x 4, 83894, 83912	2 - 4 weeks	\$540
Familial BRCA1 and BRCA2 mutations	familial breast cancer	83891, 83898, 83904, 83894, 83912	2 - 4 weeks	\$415
3 Ashkenazi BRCA1 and BRCA2 mutations	familial breast cancer	83891, 83893, 83898 x 2, 83894, 83912	2 - 4 weeks	\$450
ARX sequencing	XLAG, X-linked mental retardation, West syndrome	83891, 83898 x2, 83904 x4, 83912	4 - 6 weeks	\$1,000
ATP7B sequencing	Wilson disease	83891, 83898 x4, 83904 x9, 83912	4 - 6 weeks	\$2,025
CHD7 sequencing	CHARGE syndrome	83891, 83898 x9, 83904 x9, 83912	10 weeks	\$2,400
CDKL5/STK9 sequencing	atypical Rett syndrome	83891, 83898 x 4, 83904 x 9, 83912	4 - 6 weeks	\$2,025
GJB2 sequencing (Connexin 26)	hearing loss	83891, 83898, 83904, 83894, 83912	4 - 6 weeks	\$430
GPIIb sequencing	Bernard-Soulier syndrome	83891, 83898, 83904, 83912	4 weeks	\$400
KCNJ11 sequencing	neonatal diabetes	83891, 83898, 83904, 83912	4 weeks	\$400
LMNA sequencing (Lamin A/C)		83891, 83898 x 2, 83904 x 4, 83912	4 - 6 weeks	\$1,000



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<i>MTM1</i> sequencing	X-linked myotubular myopathy	83891, 83898 x 4, 83904 x 9, 83912	4 - 6 weeks	\$2,025
<i>NKX2.5</i> sequencing	isolated congenital heart defects	83891, 83898, 83904, 83894, 83912	4 weeks	\$430
<i>NSDHL</i> sequencing	CHILD syndrome	83891, 83898 x4, 83904 x5, 83912	4 - 6 weeks	\$1,325
Custom mutation analysis	Please call lab to discuss specific testing		4 - 6 weeks	\$390-650
MISCELLANEOUS FEES				
Culture - ALL PRENATAL DNA tests		88235-52		\$150
STAT/PRENATAL fee for known mutations, deletion/duplication testing, MPCR or UPD		99051	MAY CUT TAT IN HALF	\$200
STAT/PRENATAL fee for full gene sequencing		99051	MAY CUT TAT IN HALF	\$500
*PRENATAL: 20cc amniotic fluid, 20mgs chorionic villi, or 2 T25 flasks cultured cells, along with 3-10cc of mother's blood in an EDTA/purple top tube				