

Center for Human Genetics Laboratory, Specimen Requirements

Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Ambiguous Genitalia	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	Possible additional cells; possible FISH
Amenorrhea	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	Possible additional cells; possible FISH
Aminoglycoside Induced Hearing Loss		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Maternal Inheritance
Anemia (Chromosomes)	X				Bone Marrow	Heparinzed	2-3	2-3	2-3	Insufficient reason for referral
Anemia (Chromosomes)	X				Peripheral Blood	Heparinzed	3-5	3-5	5-7	Insufficient reason for referral
Angelman Syndrome (Chromosomes)	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (FISH)			X		Peripheral Blood	Heparinzed	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (DNA Methylation)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires phenotype
Angelman Syndrome (UPD)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires phenotype
APC (I307K)		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Autism (Chromosomes)	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	Possible DNA for Fragile X
Autism (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires concurrent chromosome study
Autistic Spectrum Disorder (Chromosomes)	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	
Autistic Spectrum Disorder (FISH)			X		Peripheral Blood	Heparinzed	2-3	3-5	5-7	FISH for dup (15)
Bcr/abl (FISH)			X		Peripheral Blood	Heparinzed	2-3	5-7	5-7	May include chromosomes
Bcr/abl (CML) (FISH)			X		Bone Marrow	Heparinzed	2-3	5-7	5-7	May include chromosomes
Bcr/abl (PCR) (Major / Minor)				X	Peripheral Blood	Heparinzed	2-3	5-7	5-7	Call Lab
Bcr/abl (PCR) (Major / Minor)				X	Peripheral Blood	EDTA / ACD-A	2-3	5-7	5-7	Call Lab
Bcr/abl [t(9;22)] (Chromosomes)	X				Bone Marrow	Heparinzed	2-3	3-5	3-5	May include FISH
Bcr/abl [t(9;22)] (Chromosomes)	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	May include FISH
Beckwith-Wiedemann (UPD 11)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Call Lab prior to sending sample
Chimerism Post-transplant		X			Peripheral Blood	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Post-transplant		X			Bone Marrow	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Pre-transplant		X			Peripheral Blood	EDTA	2-3	2-3	2-3	Pretransplant required
Chimerism Pre-transplant		X			Bone Marrow	EDTA	2-3	2-3	2-3	Pretransplant required
Chromosome Analysis	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	
Cleft Lip	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	Possible FISH #22
Connexin 26		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Cri-du-chat	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	
Cystic Fibrosis Screening		X			Peripheral Blood	EDTA	N/A	N/A	2-3	
Deep Vein Thrombosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Developmental Delay (Chromosomes)	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	
Developmental Delay (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Concurrent chromosome study highly recommended

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DiGeorge Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #22
DNA Extraction & Storage		X			Peripheral Blood	EDTA	2-3	5-7	5-7	Must have indications
Down Syndrome	X				Peripheral Blood	Heparinized	2-3	3-5	3-5	
Edwards Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Factor V HR2		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Only for Factor V Leiden Heterozygotes
Factor V Leiden Gene Mutation		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Failure to Thrive	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
FISH (not including telomeric probes)			X		Peripheral Blood	Heparinized	2-3	2-3	2-3	Must be specific
Fragile X (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Fragile X (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Chromosomes recommended
FXTAS (Fragile X tremor/ataxia syndrome)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Gonadal Dysgenesis	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Habitual Aborter	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Habitual Aborter	X				Tissue	Sterile Saline				Refrigerated
Habitual Aborter (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hearing Loss (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	3-5	5-7	
Hearing Loss (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hemochromatosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Hypotonia (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #15
Hypotonia (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Infertility (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Infertility (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Iron Overload		X			Peripheral Blood	EDTA	2-3	3-5	5-7	For hereditary hemochromatosis
Karyotype	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Klinefelter	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
LCHAD Deficiency		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Leukemia	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Lymphoblast Line (Cell Line Build Up)	(x)	(x)	(x)	(x)	Peripheral Blood	EDTA	2-3	3-5	5-7	CLBU then used for specific, designated test
MCAD		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
MELAS		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Mental Retardation (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Mental Retardation (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Concurrent chromosome study highly recommended
MERRF		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	

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Syndrome	Cytogenetics	Molecular	FISH	Sendout	Specimen Type	Media	Infant	Child	Adult	Comments
Methylation		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires Phenotype
Microdeletion Study	X		X		Peripheral Blood	Heparinized	2-3	3-5	3-5	
Miller-Dieker Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH / Possible UPD
Mitochondrial Panel		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Monosomy Study	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	Possible FISH
MTHFR Gene Mutation (C677T)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Multiple Congenital Anomalies	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Multiple Pregnancy Losses (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Multiple Pregnancy Losses (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
NARP (Leigh) Syndrome		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Transient Neonatal Diabetes (UPD 6)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Patau Syndrome	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Pellet Buildup	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
PML (rar alpha)	X		X		Peripheral Blood	Heparinized	2-3	3-5	3-5	Possible FISH #15
Prader Willi Syndrome (Chromosomes)	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Requires Phenotype
Prader Willi Syndrome (FISH)			X		Peripheral Blood	Heparinized	2-3	3-5	5-7	Requires Phenotype
Prader Willi Syndrome (DNA Methylation)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Requires Phenotype
Prader Willi Syndrome (UPD)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Premature Ovarian Failure		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
PRION Mutation Analysis		X			Peripheral Blood	DNA				Contact lab first
Prothrombin Gene Mutation (G20210A)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Pulmonary Embolism		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Retinoblastoma	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH #13
Rubenstein Taybi	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH Rubenstein region
Russel Silver (UPD 7)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Sex Chromosome abnormality	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	Possible FISH Sex chromosomes
Short Stature	X				Peripheral Blood	Heparinized	2-3	5-7	5-7	
Spongiform Encephalopathy		X			Peripheral Blood	DNA				Contact lab first
SRY (Y microdeletion)			X		Peripheral Blood	Heparinized	2-3	3-5	3-5	
t(9;22) (CML)			X		Peripheral Blood	Heparinized	2-3	5-7	5-7	
Thrombophilia		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Translocation	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	
Translocation Carrier	X				Peripheral Blood	Heparinized	2-3	2-3	2-3	

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Translocation Carrier (DNA)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	UPD for Chromosomes 6,7,14,15
Trisomy	X				Peripheral Blood	Heparinzed	2-3	2-3	2-3	
Turner Syndrome	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	
Unbalanced Translocation Carrier	X				Peripheral Blood	Heparinzed	2-3	3-5	5-7	
Uniparental Disomy		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Call Lab prior to sending sample if chr. 11 analysis
Uniparental Disomy (Amnio)		X			Cultured Cells					2 T-25 flasks at 75% confluency
Uniparental Disomy (CVS)					Cultured Cells					2 T-25 flasks at 75% confluency
UPD (14)		X			Peripheral Blood	EDTA	2-3	3-5	5-7	Rob. Translocations & Isochromosomes involving 14
Velocardiofacial Syndrome	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	Possible FISH #22, #10
Venous Thrombosis		X			Peripheral Blood	EDTA	2-3	3-5	5-7	
Williams Syndrome	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	Possible FISH #7
Wolf-Hirschorn (4p deletion)	X				Peripheral Blood	Heparinzed	2-3	5-7	5-7	Possible FISH #4p
X Inactivation		(x)		X*	Peripheral Blood	EDTA	2-3	3-5	5-7	
Y Deletion		X			Peripheral Blood	EDTA	2-3	3-5	5-7	

NOTE A Minimum of 2 ml is required for any Molecular Testing

X* Indicates Temporary Sendout because of low volumes of requested test

For more information, please contact the Lab:

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