Testing Guidelines and Best Practices

FROM THE CLINIC FOR SPECIAL CHILDREN

When to Offer Carrier Testing

- Carrier testing should be offered to both the expectant mother and her spouse; thus, samples should be sent together when possible
- Carrier testing should be offered as early as possible in the pregnancy
- Consider comprehensive carrier testing with the Plain Insight Panel™, a better value that screens for all known disease-causing DNA variants in Plain individuals

Urgent Testing

- Look for the ★ on the back of this handout for a list of our urgent tests
- For urgent tests on newborns (using cord blood), please ensure parents are lab-confirmed carriers or have had a previously affected child diagnosed by the CSC
- Ensure that the specific gene and variant (not just disease name) are documented on the requisition form as some conditions have multiple disease-causing variants

When to call the Clinic

- Call us as early as possible regarding an at-risk expectant mother so we can determine if any prenatal measures could be helpful for the baby
- Call us when a woman is in active labor and her baby is at-risk for a condition on our urgent disorder list
- Call us when a sample for an urgent test is en route to the CSC

Turnaround Times

Sample received → results available:

0-2 days: urgent targeted tests (★)
1 week: non-urgent targeted tests
4 weeks: Plain Insight Panel™

Instructions

Include when sending a sample

- Consent *and* requisition form: please use only the current version from our web site and ensure it is fully completed
- Purple top tube: labeled with the patient's first name, last name, and date of birth

Shipping reminders

- All samples should be sent at room temperature using PRIORITY OVERNIGHT shipping to: **Clinic for Special Children, Attn: Laboratory, 535 Bunker Hill Road, Strasburg, PA 17579**
- Our preferred shipper is UPS, but FedEx is suitable
- Send samples for non-urgent tests Monday-Thursday **ONLY**
- Samples drawn on weekends and/or holidays should be stored in a standard refrigerator until overnight shipping is feasible

Questions?

Call us at **717-687-9407** during normal business hours
(Monday - Friday, 9:00 AM - 5:00 PM EST)

Our latest forms can be found online
at **www.ClinicforSpecialChildren.org**

Clinic *for* Special Children®
535 Bunker Hill Road, Strasburg, PA 17579 **T** 717.687.9407 **F** 717.687.9237

Updated October 2021

Commonly Ordered Tests

Urgent Tests

Disease Name	Common Name	Variant Type	Gene	Variant
3-ß-hydroxysteroid dehydrogenase deficiency	CAH	Amish	*HSD3B2*	c.35G>A ★
Crigler-Najjar syndrome	CN1	Amish & Mennonite	*UGT1A1*	c.222C>A ★
Glutaric aciduria, type 1	GA-1	Amish	*GCDH*	c.1262C>T ★
Maple syrup urine disease	MSUD	Mennonite	*BCKDHA*	c.1312T>A ★
Ornithine transcarbomylase deficiency	OTC deficiency	Amish	*OTC*	c.422G>A ★
Pyruvate kinase deficiency	PKD	Amish	*PKLR*	c.1436G>A ★
Adenosine deaminase deficiency	SCID	Amish	*ADA*	c.646G>A ★
Severe combined immune deficiency	SCID	Amish	*RAG1*	c.2974A>G ★
Severe combined immune deficiency	SCID	Mennonite	*IL7R*	c.2T>G ★
Sudden infant death with dysgenesis of the testes	SIDDT	Amish	*TSPYL1*	c.457dupG ★
Spinal muscular atrophy	SMA	Amish & Mennonite	*SMN1*	exon 7 deletion ★
Familial hypercholanemia	Vitamin K deficiency	Amish	*TJP2*	c.143T>C ★
			BAAT	c.226A>G ★
CODAS syndrome		Amish	*LONP1*	c.2161C>G ★
Galactosemia		Amish	*GALT*	c.563A>G ★
Homocystinuria		Amish	*MTHFR*	c.1129C>T ★
Lethal neonatal rigidity and multifocal epilepsy		Amish	*BRAT1*	c.638dupA ★

Non-Urgent Tests

Hypertrophic cardiomyopathy	ANT-1	Mennonite	*SLC25A4*	c.523delC
Nephrotic syndrome, type 1	Burkholder kidney disease	Mennonite	*NPHS1*	c.1481delC
Byler disease	Byler disease	Amish	*ATP8B1*	c.923G>T
Cortical dysplasia and focal epilepsy	Casper2	Amish	*CNTNAP2*	c.3709delG
Cartilage-hair hypoplasia	CHH	Amish	*RMRP*	n.71A>G
Nemaline rod myopathy	chicken/ pigeon breast disease	Amish	*TNNT1*	c.538G>T
Non-syndromic deafness	Connexin 26	Amish & Mennonite	*GJB2*	c.35delG
Ellis-van Creveld syndrome	EVC	Amish	*EVC*	c.1886+5G>T
Susceptibility to thrombophilia	Factor 5	Amish & Mennonite	*F5*	c.1601G>A
Thyroid dyshormonogenesis 5	hypothyroidism, congenital	Amish	*DUOXA2*	c.778_779delTG
Microcephaly with chorioretinopathy	Mennonite microcephaly	Mennonite	*TUBGCP6*	c.5458T>G

Non-Urgent Tests

Disease Name	Common Name	Variant Type	Gene	Variant
Nonketotic hyperglycemia	NKH	Amish	*AMT*	c.230C>T
Nonketotic hyperglycemia	NKH	Amish	*GLDC*	c.2186delC
				c.128delA
Propionic acidemia	PA	Amish & Mennonite	*PCCB*	c.1606A>G
Phenylketonuria	PKU	Amish & Mennonite	*PAH*	c.782G>A
				c.284_286delTCA
				c.1066-11G>A
STRADA deficiency	Pretzel syndrome	Mennonite	*STRADA*	c.471-1974_1047+2194del
Severe combined immune deficiency	SCID	Mennonite	*RAG1*	c.527G>T
Nephrocerebellar syndrome	Yoder dystonia	Amish	*WDR73*	c.888delT
Aldosterone deficiency		Amish	*CYP11B2*	c.104_109delinsG
Alpha-1 antitrypsin deficiency		Amish & Mennonite	*SERPINA1*	c.1096G>A
Cardiomyopathy (dilated with arrhythmia)		Amish	*DSP*	c.699G>A
Cardiomyopathy (dilated, hypertrophic, severe neonatal)		Amish	*MYBPC3*	c.3330+2T>G
Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome		Amish	*TMCO1*	c.292_293delAG
Cystinuria		Mennonite	*SLC3A1*	c.1136+2T>C
Elliptocytosis-2/ spherocytosis		Mennonite	*SPTA1*	c.6154delG
Familial focal epilepsy and focal cortical dysplasia		Mennonite	*NPRL3*	c.349delG
Familial focal epilepsy with variable foci		Mennonite	*DEPDC5*	c.1453C>T
Familial hypercholesterolemia		Amish	*APOB*	c.10580G>A
Fragile X syndrome		Mennonite	*FMR1*	(CGG)n expansion
Gitelman syndrome		Amish	*SLC12A3*	c.1924C>G
				c.1-1471_893del
GM3 synthase deficiency		Amish	*ST3GAL5*	c.862C>T
Hereditary hemochromatosis		Amish	*HFE*	c.845G>A
Hirschsprung disease		Mennonite	*EDNRB*	c.828G>T
Limb-girdle muscular dystrophy, type 2A		Amish	*CAPN3*	c.2306G>A
Pierson syndrome		Mennonite	*LAMB2*	c.440A>G
Primary ciliary dyskinesia		Amish	*DNAH5*	c.4348C>T
Salla disease		Mennonite	*SLC17A5*	c.115C>T
Symptomatic epilepsy and skull dysplasia		Amish	*SNIP1*	c.1097A>G

★ urgent tests

(run by our laboratory staff after hours & on weekends)

Not sure which test to order?

Call us at **717-687-9407** during normal business hours

(Monday - Friday, 9:00 A.M. - 5:00 P.M. EST)

A complete list of targeted tests available through the Clinic can be found online at **www.ClinicforSpecialChildren.org**

(under the "What We Do/Diseases & Mutations" tab)