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 for both partners when possible
- Offer testing as early in the pregnancy as possible
- Consider comprehensive carrier testing for Plain couples with the Plain Insight Panel™, a better value when considering testing for three (3) or more conditions

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 confirmed carriers or had genetic testing with a previously affected child. Some conditions have multiple genetic variants associated with them, so we want to ensure we are testing for the correct variant in the baby

When to call the Clinic

- To notify us as early in the pregnancy as possible about an expectant carrier couple so we can determine if any prenatal measures could be helpful for the baby
- If a woman is in active labor and her baby is at-risk for a condition on our urgent disorder list
- When a sample for one of our urgent tests is en route to the Clinic

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 & 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 **OVERNIGHT** shipping to: **Clinic for Special Children, Attn: Laboratory, 535 Bunker Hill Road, Strasburg, PA 17579**
- 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 AM - 5 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

Commonly Ordered Tests

Disease Name	Common Name	Variant Type	Gene	Variant
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
3-ß-hydroxysteroid dehydrogenase deficiency	CAH	Amish	*HSD3B2*	c.35G>A ★
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
Crigler-Najjar syndrome	CN1	Amish & Mennonite	*UGT1A1*	c.222C>A ★
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
Glutaric aciduria, type 1	GA-1	Amish	*GCDH*	c.1262C>T ★
Thyroid dyshormonogenesis 5	hypothyroidism, congenital	Amish	DUOXA2	c.778_779delTG ★
Microcephaly with chorioretinopathy	Mennonite microcephaly	Mennonite	*TUBGCP6*	c.5458T>G
Maple syrup urine disease	MSUD	Mennonite	*BCKDHA*	c.1312T>A ★
Nonketotic hyperglycemia	NKH	Amish	*GLDC*	c.2186delC ★ c.128delA ★
Ornithine transcarbomylase deficiency	OTC deficiency	Amish	*OTC*	c.422G>A ★
Propionic acidemia	PA	Amish & Mennonite	*PCCB*	c.1606A>G
Pyruvate kinase deficiency	PKD	Amish	*PKLR*	c.1436G>A ★
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
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 ★

Disease Name	Common Name	Variant Type	Gene	Variant
Familial hypercholanemia	Vitamin K deficiency	Amish	*TJP2*	c.143T>C ★
			BAAT	c.226A>G ★
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
CODAS syndrome		Amish	*LONP1*	c.2161C>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
Galactosemia		Amish	*GALT*	c.563A>G ★
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
Homocystinuria		Amish	*MTHFR*	c.1129C>T ★
Lethal neonatal rigidity and multifocal epilepsy		Amish	*BRAT1*	c.638dupA ★
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 AM - 5 PM EST)

A complete list of targeted tests available through CSC can be found online at **www.ClinicforSpecialChildren.org (under "Our Research" tab)**