

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.14,81delC ★
Byler disease	Byler disease	Amish	ATP8B1	c.923G>T
3-β-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	∆.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 dysmorphogenesis 5	hypothyroidism, congenital	Amish	DUOX2	c.778_779delTG ★
Microcephaly with chorioretinopathy	Mennonite microcephaly	Mennonite	TUBGCP6	c.5458T>G
Maple syrup urine disease	MSUD	Mennonite	BCKDHA	c.1312T>A ★
Nonketotic hyperglycinemia	NKH	Amish	GLDC	c.2186delC ★
				c.128delA ★
Ornithine transcarbamylase 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 hypercholesterolemia	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)