Ordering Genetic Testing

Plain Insight Panel™ Carrier Screening

- **Carrier screening is recommended for all Plain couples of child-bearing age.**
- **The panel is the best value since it screens for >1,300 Plain variants in one test.**
- **Results of the panel allow for identification of couples at risk of having a child with a genetic disorder before the baby is born.**
- **The typical turnaround time is 4-6 weeks.**

The Plain Insight Panel™ (PIP) is only offered to adults 18 years or older. If possible, couples should send their blood samples at the same time. If pregnant, please indicate expected due date on requisition forms. Genetic counselors are available to explain results.

Targeted Variant Testing

- **Targeted variant testing detects single genetic variants for carrier status or diagnosis. The specific variant to be tested must be noted on the requisition form. For help determining which variant to request, contact the Clinic.**
- **Cord blood testing on newborns should only be requested if parents are confirmed carriers.**
- **The typical turnaround time is 5-7 business days for carrier screening, 1-3 business days for cord blood.**

The back of this flyer lists commonly ordered tests. Targeted variant testing for carrier status is only offered to adults 18 years or older.

Forms

ALL samples must be accompanied by fully completed consent and requisition forms.

The latest forms can be found at www.ClinicforSpecialChildren.org under 'What We Do'>Lab Services.

Shipping

Please ship samples Priority Overnight (Monday-Thursday ONLY) via UPS or FedEx to 20 Community Lane, Gordonville, PA 17529.

Samples sent via US Postal Service (USPS) should be addressed to P.O. Box 500, Intercourse, PA 17534. Receipt may be delayed.

Sample Collection

Collect ≥ 3ml of blood in a lavender top (EDTA) tube labeled with the patient's name and date of birth.

Store in a standard refrigerator until shipping. Do not freeze.

Urgent Testing Information

For urgent testing outside regular business hours, contact the Clinic to speak to the on-call provider: 717-687-9407

Any urgent testing requests will be triaged by the on-call provider based on the clinical situation and potential genetic diagnosis. Please view the reverse of this flyer for diseases that may need urgent diagnosis. Samples are to be placed in the drop box slot by our front door and couriers must call the on-call provider to confirm delivery. The turnaround time is typically 24 hours.

Clinic *for* Special Children®
Street Address | 20 Community Lane, Gordonville, PA 17529
Mailing Address | PO Box 500, Intercourse, PA 17534
T 717.687.9407 **F** 717.687.9237

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Commonly Ordered Tests

A complete list of targeted tests is available online at **www.ClinicforSpecialChildren.org**
(under the "What We Do/Diseases & Mutations" tab)

Disease Name	Common Name	Variant Type	Gene	Variant
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	CASPR2	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
Glutaric aciduria, type 1	GA-1	Amish	*GCDH*	c.1262C>T ★
Waardenburg, type 4A	Hirschsprung disease	Mennonite	*EDNRB*	c.828G>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	*AMT*	c.230C>T
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 ★
Severe combined immune deficiency	SCID	Mennonite	*RAG1*	c.527G>T ★
Severe combined immune deficiency	SCID	Mennonite	*RAG2*	c.1352G>A ★
Sudden infant death with dysgenesis of the testes	SIDDT	Amish	*TSPYL1*	c.457dupG ★

Disease Name	Common Name	Variant Type	Gene	Variant
Spinal muscular atrophy	SMA	Amish & Mennonite	*SMN1*	exon 7 deletion ★
Familial hypercholanemia	Vitamin K deficiency	Amish	*TJP2*	c.143T>C ★
			BAAT	c.226A>G ★
Galloway-Mowat (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
Cardiomyopathy		Mennonite	*SLC25A4*	c.523delC
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 c.1354C>T
Cystinuria		Mennonite	*SLC7A9*	c.544G>A c.1166C>T
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
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
Nephrotic syndrome, type 2		Amish	*NPHS2*	c.413G>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

★ these tests may require rapid diagnosis
Please contact the on-call provider to discuss a plan of action.

Not sure which test to order?

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