

Sponsored testing programs

INCREASING ACCESS TO GENETIC TESTING

METABOLIC DISORDERS AND OTHER RARE DISEASES

▶ ACUTE HEPATIC PORPHYRIAS

- For patients 16 and over who may carry a gene mutation known to be associated with the acute hepatic porphyrias
- Invitae Acute Hepatic Porphyrias Panel (ALAD, CPOX, HMBS, PPOX)
- **Invitae partner code: AHP**

▶ CHRONIC KIDNEY DISEASE

- For patients with chronic kidney disease (CKD) to facilitate diagnosis of several rare forms of CKD.
- The Invitae Progressive Renal Disease Panel includes 17 genes that are known to be associated with chronic kidney disease, including Alport syndrome, FSGS, and one form of ADPKD (PKD2).
- **Invitae partner code: CKD**

▶ HYPOPHOSPHATEMIA

- For patients aged 1 year and older who are suspected of having a genetic hypophosphatemia disorder, or who have been clinically diagnosed with X-linked hypophosphatemia (XLH)
- Invitae Hypophosphatemia Panel (13 genes)
- **Invitae partner code: XLH**

▶ INHERITED RETINAL DISEASE

- For patients suspected of having an inherited retinal disease
- The Invitae Inherited Retinal Disease panel tests approx. 250 genes for variants that are known to cause IRD
- **Invitae partner code: SPARK**

▶ LYSOSOMAL STORAGE DISEASE

- For patients suspected of having a lysosomal storage disease
- The LSDs program offers testing with 4 different panels, as well as single-gene tests
- **Invitae partner code: LYSO**

▶ PRIMARY HYPEROXALURIA

- For patients who have a family history or suspected diagnosis of primary hyperoxaluria
- Invitae Primary Hyperoxaluria Panel (AGXT, GRHPR, HOGA1) or Invitae Nephrolithiasis Panel (35 genes including AGXT, GRHPR, HOGA1)
- **Invitae partner code: PH1**

▶ SEVERE CONGENITAL NEUTROPENIA

- For individuals who may carry a genetic variant known to be associated with severe chronic, and/or congenital neutropenia, including WHIM syndrome
- The Invitae Severe Congenital Neutropenia panel analyzes 23 genes known to be associated with severe congenital neutropenia
- **Invitae partner code: PATH**

▶ UREA CYCLE DISORDERS

- For patients suspected of having a urea cycle disorder
- The Invitae Hyperammonemia Panel analyzes genes that are associated with the enzymes and transporter proteins responsible for the production and detoxification of ammonia, the waste product of protein metabolism
- **Invitae partner code: UCD**

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CARDIOLOGY AND NEUROLOGY

► CARDIOMYOPATHY AND ARRHYTHMIA

- For patients suspected of having a familial cardiomyopathy or arrhythmia
- This program offers 3 different panel options for inherited cardiomyopathies and arrhythmias
- **Invitae partner code: CARDIO**

► HEREDITARY ATTR AMYLOIDOSIS

- For adults 18 and over who may carry a gene mutation known to be associated with hereditary ATTR (hATTR) amyloidosis
- Invitae Cardiomyopathy Comprehensive Panel (~50 genes), Invitae Comprehensive Neuropathies Panel (~70 genes), or Invitae Transthyretin Amyloidosis Test (TTR)
- **Invitae partner code: TTR**

► MUSCULAR DYSTROPHY

- For patients suspected of having a muscular dystrophy
- Invitae Dystrophinopathies Test (DMD gene), Invitae Limb-Girdle Muscular Dystrophy Panel (~35 genes), Invitae Comprehensive Muscular Dystrophy Panel (~55 genes), or Invitae Comprehensive Neuromuscular Disorders Panel (~120 genes)
- **Invitae partner code: MDYS**

► PEDIATRIC EPILEPSY

- For children up to 60 months old who have experienced an unprovoked seizure
- Invitae Epilepsy Panel (~180 genes)
- **Invitae partner code: Behind the Seizure**

► PERIODIC PARALYSIS

- For adults 18 and over who experience episodic muscle weakness or temporary paralysis provoked by common triggers for primary hyperkalemic or hypokalemic periodic paralysis
- Invitae Periodic Paralysis Panel (SCN4A, CACNA1S, KCNJ2, RYR1)
- **Invitae partner code: UPP**

► SPINAL MUSCULAR ATROPHY (SMA)

- To help in the diagnosis of SMA or carrier status identification of SMA
- Invitae SMA Panel (SMN1, SMN2), Invitae SMA STAT Panel (results provided within 4 days from sample accessioning), or Invitae SMA Carrier Screen (SMN1 only)
- **Invitae partner code: SMA**

To learn more, please visit www.invitae.com/sponsored-testing

* Sponsored testing for AHP, Behind the Seizure, CARDIO, LYSO, MDYS, PH1, and TTR is available in the US and Canada; all other programs are available only in the US. For more information and to see if your patient meets eligibility criteria, visit www.invitae.com/sponsored-testing.