# Sponsored testing programs

## INCREASING ACCESS TO GENETIC TESTING

INVITAE

## 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
- The Invitae Acute Hepatic Porphyrias Panel (4 genes)
- **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 6 months 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**

### ▶ SKELETAL DYSPLASIA

- For patients with signs or symptoms suggestive of or consistent with a diagnosis of skeletal dysplasia
- The Invitae Expanded Skeletal Dysplasia Panel (109 genes)
- **Invitae partner code: SKEL**

### ▶ 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**

**HEADQUARTERS** | 1400 16th Street, San Francisco, CA 94103 | **ONLINE** | www.invitae.com | **CONTACT** | www.invitae.com/contact

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## CARDIOLOGY AND NEUROLOGY

### ▶ CARDIOMYOPATHY AND ARRHYTHMIA

- For patients suspected of having a familial cardiomyopathy or arrhythmia
- This program uses the Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel (up to 150 genes)
- **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 any child under the age of 8 who has had an unprovoked seizure.
- Invitae Epilepsy Panel (~190 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 (CACNA1S, KCNJ2, RYR1, SCN4A)
- **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**

** Sponsored testing for AHP, Behind the Seizure, CARDIO, LYSO, MDYS, PH1, PATH, 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.*

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