INVITAE

PTC THERAPEUTICS™

PTC PINPOINT™
CP SPECTRUM GENETIC TESTING PROGRAM

PTC Pinpoint™ CP Spectrum Program

PROGRAM DESCRIPTION

Invitae and PTC Therapeutics have partnered to offer the PTC Pinpoint™ CP Spectrum sponsored testing program, which breaks down many barriers to genetic testing for cerebral palsy (CP). Individuals with symptoms suggestive of CP in the absence of risk factors for an acquired brain injury are eligible for the program. These individuals have access to no-charge genetic testing and counseling to help identify an underlying genetic etiology, such as Aromatic L-amino Acid Decarboxylase (AADC) deficiency, which may be amenable to specific medical management or treatment options.

ABOUT CEREBRAL PALSY SPECTRUM

CP is the most common cause of motor disability in children with an estimated incidence of 2 to 3 per 1,000 live births in the U.S.[1,2]

The term cerebral palsy represents a heterogeneous group of neurodevelopmental conditions characterized by impairment of movement, posture, and motor function[2] with an onset of symptoms typically before 2 years of age.[3]

Identifying an underlying genetic etiology for CP may impact clinical management and help reveal specific disease-modifying treatment.[4]

SIGNS AND SYMPTOMS THAT ARE SUGGESTIVE OF AN UNDERLYING GENETIC ETIOLOGY FOR CP INCLUDE:

- No documented perinatal risk for brain injury
- Full term birth
- Brain MRI that is normal or inconsistent with acquired brain injury
- Onset of abnormal motor symptoms after a period of normal development
- Progressive neurologic symptoms
- Fluctuation of muscle tone or motor symptoms
- Worsening of neurologic symptoms with fever and/or illness
- Hypotonia as an isolated symptom
- Ataxia as a prominent feature
- Evidence of peripheral neuromuscular disease
- Abnormal eye movements
- Family history of similarly affected individuals

PANEL INFORMATION

The PTC Pinpoint™ CP Spectrum program offers testing with the Invitae Cerebral Palsy Spectrum Disorders Panel, which analyzes a broad panel of 265 genes to determine the underlying etiology of cerebral palsy (CP). Once Invitae receives your patient's blood, buccal, or saliva, their results will be available in 10 to 21 calendar days, on average.

GENETIC COUNSELING

Individuals tested through the PTC Pinpoint™ CP Spectrum Program are eligible for post-test genetic counseling to help them understand their test results. This service is provided through GeneMatters, a third-party genetic counseling service, and is made available by Invitae at no charge as part of the program. To schedule a genetic counseling appointment, please contact GeneMatters at 1-866-741-5331 or schedule online at www.gene-matters.com (use partner code: **CPSD**).

FAMILY VARIANT TESTING

Invitae offers family variant testing for all blood relatives of patients who undergo testing at Invitae and are found to have a pathogenic or likely pathogenic variant. Our family variant testing reports on the variant(s) that was identified in the original family member, as well as any other pathogenic variant(s) found in the full gene.

If your patient tests positive through the PTC Pinpoint™ CP Spectrum Program, all of his or her blood relatives are eligible for family variant testing.

For more information or to order a test,
please visit www.invitae.com/ptc-pinpoint-cp-spectrum/

While third parties and commercial organizations may provide financial support for this program, tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing and counseling is available in the US and Canada only. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use, or support any other products or services from Invitae or from third parties or commercial organizations.

Reference:

1. Lee RW, Poretti A, Cohen JS, et al. A diagnostic approach for cerebral palsy in the genomic era. Neuromolecular Med. 2014;16(4):821-844. doi:10.1007/s12017-014-8331-9.
2. van Eyk CL, Corbett MA, Maclennan AH. The emerging genetic landscape of cerebral palsy. Handb Clin Neurol. 2018;147:331-342. doi:10.1016/B978-0-444-63233-3.00022-1.
3. Pearson TS, Pons R, Ghaoui R, Sue CM. Genetic mimics of cerebral palsy. Mov Disord. 2019;34(5):625-636. doi:10.1002/mds.27655.
4. Zarrei M, Fehlings DL, Mawjee K, et al. De novo and rare inherited copy-number variations in the hemiplegic form of cerebral palsy. Genetics in Medicine : Official Journal of the American College of Medical Genetics. 2018 Feb;20(2):172-180. DOI: 10.1038/gim.2017.83.