INVITAE

PTC THERAPEUTICS

PTC PINPOINT™
NEUROTRANSMITTER DISORDERS PROGRAM

PTC Pinpoint™ Neurotransmitter Disorders Program

PROGRAM DESCRIPTION

Invitae and PTC Therapeutics have partnered to offer the PTC Pinpoint™ Neurotransmitter Disorders sponsored testing program, which provides no-charge genetic testing and counseling for individuals in the U.S. and Canada suspected of having a neurotransmitter disorder.

ABOUT NEUROTRANSMITTER DISORDERS

The inborn errors of neurotransmission are a genetically heterogeneous set of conditions affecting the metabolism and transport of neurotransmitters.[1,2,3]

These disorders typically present in infancy or childhood, but later onset presentations have also been described.[1,2]

Treatments and clinical trials are available for some of these disorders so early diagnosis may help improve long term outcomes.[4,5]

To biochemically diagnose many neurotransmitter disorders, neurotransmitter levels must be obtained by extracting cerebral spinal fluid (CSF). **In such cases, molecular testing may help avoid a lumbar puncture and the associated risks for the patient.**

PANEL INFORMATION

The PTC Pinpoint™ Neurotransmitter Disorders program offers testing with the Invitae Neurotransmitter Disorders Panel, which analyzes up to 45 genes that are associated with disorders of monoamine metabolism, GABA metabolism, and neurotransmitter receptors and transporters. Once Invitae receives your patient's blood, buccal, or saliva specimen, their results will be available in 10 to 21 calendar days, on average.

GENETIC COUNSELING

Individuals tested through the PTC Pinpoint™ Neurotransmitter Disorders 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: PTC).

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™ Neurotransmitter Disorders 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

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.

References

1. Ng J, Papandreou A, Heales SJ, Kurian MA. Monoamine neurotransmitter disorders–clinical advances and future perspectives. Nat Rev Neurol. 2015 Oct;11(10):567-84. doi: 10.1038/nrneurol.2015.172. Epub 2015 Sep 22. PMID: 26392380.
2. Mercimek-Mahmutoglu S, Sidky S, Hyland K, Patel J, Donner EJ, Logan W, Mendoza-Londono R, Moharir M, Raiman J, Schulze A, Siriwardena K, Yoon G, Kyriakopoulou L. Prevalence of inherited neurotransmitter disorders in patients with movement disorders and epilepsy: a retrospective cohort study. Orphanet J Rare Dis. 2015 Feb 8;10:12. doi: 10.1186/s13023-015-0234-9. PMID: 25758715; PMCID: PMC4342151.
3. García-Cazorla A, Artuch R, Gibson KM. Inborn metabolic diseases: diagnosis and treatment. 6th ed. Heidelberg: Springer; 2016. Chapter 29, Disorders of Neurotransmission; p. 415–427.
4. Ng J, Heales SJ, Kurian MA. Clinical features and pharmacotherapy of childhood monoamine neurotransmitter disorders. Paediatr Drugs. 2014 Aug;16(4):275-91. doi: 10.1007 s40272-014-0079-z. PMID: 25011953; PMCID: PMC4102824.
5. Kurian MA, Gissen P, Smith M, Heales S Jr, Clayton PT. The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurol. 2011 Aug;10(8):721-33. doi: 10.1016/S1474-4422(11)70141-7. PMID: 21777827.

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