

PATHOLOGY & LAB MEDICINE Diagnostic Labs

Genomic Diagnostics @ CHOP



Diagnostic accuracy of pediatric CNS tumors is complicated. The integration of molecular and pathological findings, however, can play a key role in assessing tumor types, likelihood of responsiveness to treatment, and monitoring for recurrence in some scenarios.

The cancer genomic testing at CHOP offers comprehensive cytogenetic and molecular testing which facilitates diagnosis, prognosis, monitoring and information on germline cancer risk assessment.

Cancer Panels	Forms/Specimen Handling
Comprehensive Somatic NGS Panels (Solid+Fusion)	
Cancer Transcriptome Analysis (RNA Sequencing)	
Paired Tumor-Normal Sequencing Panels	
Hereditary Cancer Panels	
CNS Methylation	For CNS Methylation Req Form, please contact clinicallyaboutreach@chop.edu

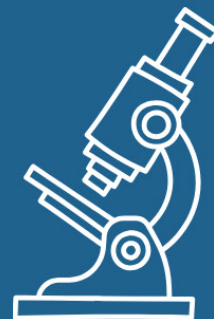


Children's Hospital
of Philadelphia®
Pathology &
Laboratory Medicine

PATHOLOGY & LAB MEDICINE

Diagnostic Labs

Neuropathology @ CHOP



Pediatric CNS tumor histology and biology has unique features compared to adults and requires pediatric expertise to accurately diagnose. The specific type and grade of a tumor, determined through neuropathology, can provide important prognostic information, guiding treatment strategies and predicting outcomes.

As the largest group of working pediatric neuropathologists anywhere in the US, CHOP has renowned expertise in the areas of pediatric brain tumors. With a wide range of consultation services, these evaluations can be requested by outside clients.

Consultative Services	Forms/Specimen Handling
Brain biopsies (tumors and non-neoplastic disease)	
Autopsy brain and spinal cord examinations	



**Children's Hospital
of Philadelphia®**
Pathology &
Laboratory Medicine

LIQUID BIOPSY

NEUROBLASTOMA PANEL

(CIRCULATING TUMOR DNA)



Children's Hospital
of Philadelphia®
Division of Genomic Diagnostics

PEDIATRIC CANCER DIAGNOSTICS

Co-designed by leading experts in somatic diagnostics and pediatric clinical oncology

Overview	Assay	Clinical Utility
This customized panel utilizes Next Generation Sequencing (NGS) to detect and monitor tumor-derived genetic variants associated with neuroblastoma.	This assay provides sequence analysis of 23 genes associated with neuroblastoma, along with amplification status of <i>MYCN</i> .	Pediatric tumor biology is unique and requires pediatric-focused somatic diagnostics to facilitate disease monitoring, risk stratification, and therapeutic decision-making.

Regions of Interest (ROI) (Hg19): *ALK*, *ARID1A*, *ARID1B*, *ATRX*, *BRAF*, *CDK4*, *CDKN2A*, *CDKN2B*, *CIC*, *ERBB2*, *FGFR1* (NM_023110, exons 9–19), *HRAS*, *KRAS*, *MDM2*, *MET*, *MYCN* (NM_005378, partial exon 3 for copy number analysis only), *NF1*, *NRAS*, *PHOX2B*, *PTPN11*, *SMARCA4* (NM_003072 exons 17–20, 25 and 26), *TERT* (NM_198253, promoter region), and *TP53*. All exons of the genes listed and the flanking intronic sequences are included unless otherwise specified.

COLLECTION INFO	KIT REQUEST	TEST INFO
Sample: Peripheral Blood Collection: 2 cfDNA STRECK tubes Performed: M–Th 9–4pm EST TAT: 14 days CPT code: 81462		

Ensuring children everywhere lead healthier lives		
Genomic Diagnostic Lab	Solid Tumor Program	Laboratory Outreach
The GDL offers state-of-the-art testing with experienced scientists and a comprehensive menu for pediatric constitutional and somatic conditions.	The Solid Tumor Program is one of the world’s leading treatment centers for high-risk and relapsed neuroblastoma with a treatment plan unique to each child.	The lab outreach program provides outside partners access to CHOP’s extensive test menu to address the diagnostic complexities of pediatric conditions worldwide.
For clinical and assay questions: DGDGeneticCounselor@chop.edu	For referrals and 2nd opinion questions: 267-426-0762	For outreach-related inquiries: ClinicalLabOutreach@chop.edu