CONNECTGENE
Genomics Oncology

Unlock the Power of Genomics

KIDS CANCER TARGETED THERAPY ASSAY

Cancer in children and young adults is different to cancers in adults. The types of cancers and the driving genetic mutations behind the cancers differ to those typically seen in adults.

The Kids Cancer Targeted Therapy Assay has been developed by leading researchers and paediatric oncologists to comprehensively target the genetic drivers of childhood cancers.

Understanding the genetic mutations behind a child's cancer is the key to an accurate diagnosis. Having an accurate diagnosis prior to starting treatment allows for a targeted and personalised treatment specific to your child's cancer to be used right from the start of their diagnosis, helping to optimise treatment outcomes.

What is the Kids Cancer Targeted Therapy Assay?

The Kids Cancer Targeted Therapy Assay is the first test designed for targeting multiple forms of childhood cancer. It is a specifically designed, targeted but comprehensive panel allowing detection of genetic mutations in a range of childhood cancers

The genes covered on this assay are commonly implicated in a range of childhood cancers, including leukemias, brain tumours, sarcomas, and embryonal tumours, including neuroblastoma, retinoblastoma, Wilm's tumour, and liver tumours.

How could this benefit my child?

Cancer is a complex disease, caused by a wide range of genetic mutations that can be present in many combinations. Each tumour has a unique genetic makeup. Genomic testing can be used to analyse the DNA of a tumour and may identify the genetic mutations that are unique to that cancer. Paediatric cancers, such as soft-tissue tumours, are often difficult to diagnose by conventional diagnostic methods, leading to an incomplete or inaccurate diagnosis. Genomic testing provides a powerful diagnostic tool, and every cancer patient deserves an accurate diagnosis.

The Kids Cancer Targeted Therapy Assay may identify the specific mutations unique to your child's cancer. Once we have identified the mutations, we may find targeted solutions to help treat them.

Targeted therapy has been identified for tumours with *NTRK* gene fusions that are found in a small number of paediatric tumours and there are a number of gene mutations that occur in neuroblastoma, embryonal tumours, Langerhans cell histiocytosis, Ewing sarcoma, sarcomas and leukaemia that have potential targeted therapies. A number of paediatric precision oncology trials are underway and this will improve our understanding of the clinical impact of targeted therapies.

Genomic testing can also help identify missed or unexpected genetic mutations that were not identified by conventional testing methods, which may also highlight potential matched targeted therapies

Investing in genomic testing, to obtain a complete diagnosis and to select appropriate therapy is a small cost compared with the money and time that may be wasted on ill-chosen therapies. By identifying potential targetable mutations early on in your child's diagnosis, targeted therapies can be used where they are most likely to be effective.

Genomic testing provides a personalised approach to cancer treatment, giving your child their best chance of survival. Clinical trials of over 70,000 patients have shown that personalised therapy, based on genomic profiling of tumours, is the most effective way to improve outcome, with higher response rates, longer progression free and overall survival, and fewer deaths related to toxic effects across cancers. [3-5]

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What does the Kids Cancer Targeted Therapy Assay cover?

This assay covers genes commonly implicated in a range of childhood cancer types, including leukemias, brain tumours, sarcomas, neuroblastomas, retinoblastomas, Wilm's tumour, and liver tumours.

The Kids Cancer Targeted Therapy Assay includes a large 97-gene translocation/fusion panel (>1700 fusion isophorme variants), as gene fusions and copy number alterations are more common in childhood cancers compared to adult cancers. The Assay also includes an 82-DNA target panel with comprehensive coverage of all relevant mutations, 44 targets with full exon coverage, specifically tumour suppressor genes, and 24 CNV targets, including several key driver DNA mutations that have been described in different paediatric tumors - SMARCB1 mutations in rhabdoid tumours, ALK point mutations in neuroblastomas, and BRAF point mutations in paediatric gliomas. The genes are all covered by the Kids Cancer Targeted Therapy Assay

Comprehensive mutation coverage (86)			CNV (28)	Full exon coverage (44)		Fusion and expression (97)			
ABL1	*FASLG*	*MYC*	*ABL2*	*APC*	*PTEN*	*ABL1*	*JAK2*	*NTRK1*	*TAL1*
ABL2	*FBXW7*	*MYCN*	*ALK*	*ARID1A*	*RB1*	*ABL2*	*KAT6A*	*NTRK2*	*TCF3*
ALK	*FGFR1*	*NCOR2*	*BRAF*	*ARID1B*	*RUNX1*	*AFF3*	*KMT2A*	*NTRK3*	*TFE3*
ACVR1	*FGFR2*	*NOTCH1*	*CCND1*	*ATRX*	*SMARCA4*	*ALK*	*KMT2B*	*NUP214*	*TP63*
AKT1	*FGFR3*	*NPM1*	*CDK4*	*CDKN2A*	*SMARCB1*	*BCL11B*	*KMT2C*	*NUP98*	*TSLP*
ASXL1	*FLT3*	*NRAS*	*CDK6*	*CDKN2B*	*SOCS2*	*BCOR*	*KMT2D*	*NUTM1*	*TSPAN4*
ASXL2	*GATA2*	*NT5C2*	*EGFR*	*CEBPA*	*SUFU*	*BCR*	*LMO2*	*NUTM2B*	*UBTF*
BRAF	*GNA11*	*PAX5*	*ERBB2*	*CHD7*	*SUZ12*	*BRAF*	*MAML2*	*PAX3*	*USP6*
CALR	*GNAQ*	*PDGFRA*	*ERBB3*	*CRLF1*	*TCF3*	*CAMTA1*	*MAN2B1*	*PAX5*	*WHSC1*
CBL	*H3F3A*	*PDGFRB*	*FGFR1*	*DDX3X*	*TET2*	*CCND1*	*MECOM*	*PAX7*	*YAP1*
CCND1	*HDAC9*	*PIK3CA*	*FGFR2*	*DICER1*	*TP53*	*CIC*	*MEF2D*	*PDGFB*	*ZMYND11*
CCND3	*HIST1H3B*	*PIK3R1*	*FGFR3*	*EBF1*	*TSC1*	*CREBBP*	*MET*	*PDGFRA*	*ZNF384*
CCR5	*HRAS*	*PPM1D*	*FGFR4*	*EED*	*TSC2*	*CRLF2*	*MKL1*	*PDGFRB*	
CDK4	*IDH1*	*PTPN11*	*GLI1*	*FAS*	*WHSC1*	*CSF1R*	*MLLT10*	*PLAG1*	**Gene Expression**
CIC	*IDH2*	*RAF1*	*GLI2*	*GATA1*	*WT1*	*DUSP22*	*MN1*	*RAF1*	
CREBBP	*IL7R*	*RET*	*IGF1R*	*GATA3*	*XIAP*	*EGFR*	*MYB*	*RANBP17*	*BCL2*
CRLF2	*JAK1*	*RHOA*	*JAK1*	*GNA13*		*ETV6*	*MYBL1*	*RARA*	*BCL6*
CSF1R	*JAK2*	*SETBP1*	*JAK2*	*ID3*		*EWSR1*	*MYH11*	*RECK*	*wFGFR1*
CSF3R	*JAK3*	*SETD2*	*JAK3*	*IKZF1*		*FGFR1*	*MYH9*	*RELA*	*FGFR4*
CTNNB1	*KDM4C*	*SH2B3*	*KIT*	*KDM6A*		*FGFR2*	*NCOA2*	*RET*	*IGF1R*
DAXX	*KDR*	*SH2D1A*	*KRAS*	*KMT2D*		*FGFR3*	*NCOR1*	*ROS1*	*MET*
DNMT3A	*KIT*	*SMO*	*MDM2*	*MYOD1*		*FLT3*	*NOTCH1*	*RUNX1*	*MYCN*
EGFR	*KRAS*	*STAT3*	*MDM4*	*NF1*		*FOSB*	*NOTCH2*	*SS18*	*MYC*
EP300	*MAP2K1*	*STAT5B*	*MET*	*NF2*		*FUS*	*NOTCH4*	*SSBP2*	*TOP2A*
ERBB2	*MAP2K2*	*TERT*	*MYC*	*PHF6*		*GLI1*	*NPM1*	*STAG2*	
ERBB3	*MET*	*TPMT*	*MYCN*	*PRPS1*		*GLIS2*	*NR4A3*	*STAT6*	
ERBB4	*MPL*	*USP7*	*PDGFRA*	*PSMB5*		*HMGA2*			
ESR1	*MSH6*	*ZMYM3*	*PIK3CA*	*PTCH1*					
EZH2	*MTOR*								

How do I organise testing?

CG Genomics Oncology aims to educate patients and their families on their cancer type and empower them with the knowledge to take control of their treatment plans. As each patient's case is unique, there is no "one size fits all" when it comes to testing. We encourage you to contact CG Genomics Oncology, and we can work with you and your oncologist/ specialist, to determine what tests would benefit you.