

# Cancer Biology Newsletter

Issue #3 | October 2025





The aim of the Cancer Biology research theme is to better understand the biology of pediatric cancers and accelerate research efforts by removing barriers to collaboration, increasing research pathways, and building infrastructure to enable knowledge, expertise, and data sharing across Canada.

Learn more here: <u>Cancer Biology</u>



# Cancer Biology Theme

IN THIS ISSUE: Learn about the exciting advances in liquid biopsy testing from Cancer Biology Theme stakeholders Vincent-Philippe Lavallée and Chantal Richer. We also feature project updates and take an in-depth look at our Theme's newest approved initiative — the ACCESS National Biobanking Network.

**GOOD NEWS:** The Canadian Institutes of Health Research has <u>approved ACCESS' request for a no-cost extension</u>. This means projects will be eligible to complete their deliverables by <u>March 31, 2027</u> (previously March 31, 2026).

#### **CONTENT OVERVIEW:**

- O4-05 <u>Featured Article:</u> Liquid Biopsies in Pediatric Oncology: A Promising Breakthrough in Quebec Led by CHU Sainte-Justine
- 06-07 <u>Featured Project:</u> ACCESS National Biobanking Network
- 08-13 Our Projects
  - 14 Events & Opportunities
- 15-16 Glossary

# **Featured Article**

#### <u>Liquid Biopsies</u> in Pediatric **Oncology: A Promising Breakthrough in Quebec Led** by CHU Sainte-Justine

Article by Manon Nayrac

In a context where next-generation sequencing technologies have revolutionized diagnostics in pediatric oncology, one major challenge remains: access to tumour tissue. Some To tailor this approach to the realities of tumours are too small to provide a sufficient pediatric care, Chantal Richer optimized a lowsample, others are inaccessible, or their biopsy pass whole-genome sequencing (WGS) poses significant clinical risks. Faced with these method, already used clinically for nonlimitations, Vincent-Philippe Lavallée, pediatric invasive prenatal testing of trisomy from hemato-oncologist, and Chantal Richer, circulating fetal DNA. "We worked to align our research assistant at Centre de recherche protocol with existing practices at CHU Sainte-Azrieli du CHU Sainte-Justine, set out to Justine to ensure smooth clinical adoption and develop an innovative solution: sequencing of avoid unnecessary regulatory hurdles," she liquid biopsies.

Since January 2024, their team has been working to implement the analysis of circulating tumour DNA (ctDNA) in blood, a less invasive approach that still yields rich molecular insights.

"This strategy not only compensates for the lack of tissue biopsy but also captures tumour heterogeneity that is often missed by conventional sampling. In pediatrics, every milliliter of blood counts. Optimizing protocols is therefore critical," explains Vincent-Philippe.

Two sequencing platforms were considered: Illumina, already certified for clinical use at CHU Sainte-Justine, and Oxford Nanopore offers Technologies, which promising capabilities for longitudinal monitoring. To enable rapid clinical integration, the team chose Illumina, while maintaining a long-term vision for the project. Indeed, beyond the initial diagnosis, liquid biopsies have the potential to transform patient follow-up by enabling early relapse detection and real-time treatment adjustments.

explains.

Vincent-Philippe Lavallée is a hemato-oncologist at CHU Sainte-Justine and a researcher at the Centre de recherche Azrieli du CHU Sainte-Justine (CRA-CHUSJ). His research focuses on leukemia genomics, bulk and single-cell sequencing, and computational biology.

Chantal Richer is a research assistant at CRA-CHUSJ with an expertise in transcriptomics. genomics, and sequencing data analysis.

The ambition of Vincent-Philippe, Chantal, and their team is clear: to develop a robust, harmonized, and accessible test that can be integrated into provincial clinical practice and serve as a model for other jurisdictions. While the

path to clinical validation is complex, requiring protocols, certifications, and proof of feasibility, their previous experience with the 2023 whole implementation of exome transcriptome sequencing in clinical care for all

and

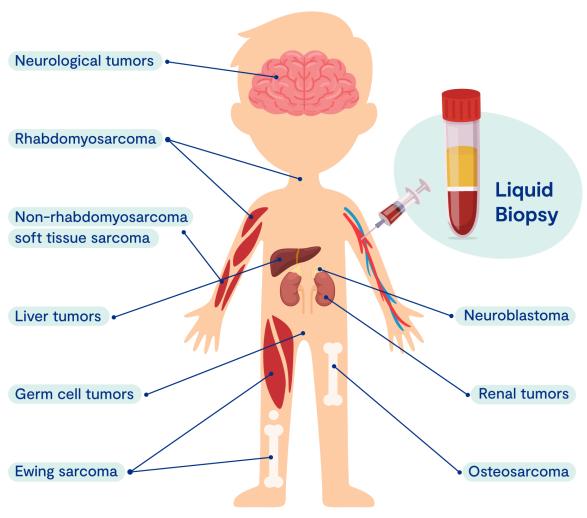


Image provided by V-P Lavallée

children in Quebec, supported by the Ministry of Health and Social Services, gives them a head start.

The national ACCESS initiative provides a dynamic platform for the development of several innovative projects that leverage circulating biomarkers, including DNA, RNA, proteins and methylation profiles, to enhance diagnostics and deepen our understanding of disease biology.

Liquid biopsy research is a key component of this broader effort, supported through various ACCESS-led initiatives, such as:

- The rapid integration of liquid biopsy testing into clinical laboratories across Canada. through the Molecular Pathology Strategy led by Drs. Nobre and Lange, and
- The exploration of pediatric cancer biology through cutting-edge studies, including the Sarcoma MetAstasis Research Taskforce (SMART) led by Drs. Garzia, Gladdy, and Sorensen, and Deciphering 3D chromatin states permissive to driver alterations in High-Grade Childhood Brain Tumours, led by Drs. Jabado and Ramaswamy.

This project embodies a modern vision of pediatric precision medicine: less invasive, faster, and better aligned with clinical realities. Most importantly, it paves the way for more equitable access to molecular diagnostics for all children with cancer.

# Featured Project

### National Research Platform: Biobanking

**Project Name:** ACCESS National Biobanking Network

**Project Leads:** Jonathan Bush & Jennifer Chan

#### **Brief Description**

Biobank infrastructure is essential advancing research, especially in rare diseases, by providing organized access to biological samples and clinical information. Various international biobanks exist that support this type of research. In Canada, several initiatives exist to improve pediatric, adolescent, and young adult (AYA) cancer research, including projects like the PRecision Oncology For Young peopLE (PROFYLE) and the Marathon of Hope Network. However, many pediatric and AYA cancer researchers are not fully utilizing these resources. potentially due to limited visibility, funding, and support, as well as complicated agreements between institutions.

A strong, accessible national biobanking system would greatly benefit cancer research by enabling studies of rare cancers, capturing diverse Canadian demographics, fostering collaboration across research initiatives, and opening new research avenues in policy, public health, and clinical practice.

#### Aims

We aim to create a national pediatric and AYA oncology biobanking network bringing together biobankers, researchers, People With Lived Experience (PWLE), and other partners across Canada. This initiative will focus on strengthening collaboration in biobanking by developing shared guidelines and launching a virtual platform to showcase and provide access to biospecimens for research. Together, these efforts will make samples easier to find and use, helping to advance pediatric cancer research nationwide.

We will also support projects that respond to the needs of the biobanking community, as identified through a national needs assessment survey.

#### **Key Deliverables**

- Establish a collaborative National Biobanking Network and Network Governance Committee
- Establish the ACCESS Virtual Biobanking Platform - a common inventory online search (and request) platform
- Provide guidelines and standard operating procedures to support biobanks and biobank users
- Support biobanking projects requiring collaboration, larger scale resources, or national coordination that are identified through a needs assessment survey.



#### **Achievements**

The proposal funding this project was approved by ACCESS in July of 2025. As the ACCESS National Biobanking Network builds and extends on the earlier work of the PROFYLE Biobanking Node, the project team is well-informed and coordinated to get the work underway. Additionally, 3 PWLE from the pediatric cancer community have joined the network.

#### **Current Focus**

The Biobanking Network has re-launched its regular meetings, which currently take place on the fourth Wednesday of each month at 1:00 p.m. ET, with the next meeting scheduled on November 29th.

We are actively identifying and engaging Canadian biobanks that collect samples from strengthening connections and collaboration across the country.

A draft Terms of Reference for the Biobanking Network Governance Committee has been written, and membership is currently being finalized. In parallel, we have identified individuals and potential solutions for several key roles within the Network, including the Project Coordinator, Needs Assessment Survey Coordinator, and Database Administrator.

Work is also underway to design a needs assessment survey, guided by responses from our initial questionnaire, to better understand and address the priorities of the biobanking community. Finally, the team is refining the list of data elements that will be featured in the upcoming virtual inventory search platform, a key step in evaluating and selecting a suitable platform for the Network's needs.

#### **Requests and Opportunities**

We are seeking Canadian biobanks that bank biospecimens from children and AYA with cancer to display biospecimen details on a virtual platform. The network will provide much of the support required, there is no up-front commitment to participate, and biobanks will have control over what information is displayed.

We invite all biobanking leaders, researchers, and individuals interested in our activities to join our monthly meetings.

#### pediatric and AYA cancer patients, with the goal of What is a virtual biobanking platform and what impact would it have on research?

A virtual biobanking platform is an online system that connects multiple physical biobanks through a centralized, searchable database. It does not store biological samples itself, but rather provides a unified view of where and what types of samples exist across different institutions.

Currently, investigators need to contact each biobank individually to find relevant samples—a process that can be time-consuming and inefficient. The proposed virtual platform would allow all participating pediatric and AYA biobanks in Canada to summarize their available participant samples in one place.

Through this system, researchers could search a single, secure site for de-identified samples using standardized data labels, making it much easier to identify what is available across the network. Regular updates from participating biobanks would ensure that the platform's inventory remains current and accurate.

Ultimately, this virtual biobank would streamline access to valuable biological materials, reduce administrative and logistical barriers, and enable more timely and collaborative research across pediatric and AYA cancer studies. This would accelerate discovery and improve the ability of researchers to address critical questions in these populations.

# Our Projects

#### National Research Project: Leukemia

**Project Name:** Diagnostic and precision intervention for leukemia arising in infancy

Project Lead: Sonia Cellot

#### **Brief Description**

Recent advancements in genetics have revealed that pediatric leukemia is much more diverse than we previously thought, but this diversity is just starting to be considered in how we diagnose, treat, and monitor the disease. Our goal is to create a national team of experts to study leukemia arising in infancy and accelerate research to improve treatments and outcomes. Our goal is to eventually apply this as a model to other high-risk leukemia subtypes.

#### **Recent Achievements**

- Identified a previously unrecognized subtype of leukemia that arises in infancy linked to the discovery and validation of a new <u>fusion gene</u>.
- Curated a list of drugs that will be used for <u>pharmacotyping</u> of patient samples.
- Sequenced 4 cord blood samples by single cell <u>RNASeq</u> at Centre Hospitalier Universitaire Ste-Justine. 4 more will be processed at the BC Children's Hospital to build a reference atlas for leukemia sample analyses.

#### **Current Focus**

Our focus is on performing <u>drug screens</u> of priority samples; generating <u>patient-derived xenografts (PDXs)</u> from priority samples; and performing single cell RNASeq on priority samples.

#### **Requests & Opportunities**

We are looking for a Person With Lived Experience to join the project team.

For more details, please visit the ACCESS website or <u>first issue</u> of our newsletter.



# Our Projects

## National Research Project: Brain Tumour

Project Name: Deciphering 3D <u>chromatin</u> states permissive to <u>driver alterations</u> in High-Grade Childhood Brain Tumours

**Project Co-Leads:** Nada Jabado & Vijay Ramaswamy

#### **Brief Description**

High-grade brain tumours have distinct genetic alterations and changes in the way DNA is organized (chromatin architecture) at the core of the processes that sustain tumour formation. This project examines how 3D chromatin changes structure and function over time (4D) and how tumour cells communicate with each other and with their surrounding environment and aims to identify new therapeutic targets.

#### **Recent Achievements**

We analyzed detailed single-cell data from high-grade brain tumours (diffuse midline gliomas and posterior fossa group A ependymomas) that carry oncohistone mutations. These are changes (mutations) in a special kind of protein called a histone, which helps organize and control how DNA is packed and used inside cells. From this, we found changes in the structure of the DNA and copy numbers in individual cells and used that information to group the cells into subpopulations that likely came from the same original cancer



cell. We also discovered that the 3D organization of the DNA in these tumours still closely resembles their cell of origin, which may help us find new treatment targets. We're continuing to explore these and other datasets for more insights.

To study how DNA is organized and active in individual tumour cells and in their surrounding environment, we used advanced mapping tools called 10X Visium HD and Xenium. This

approach, called <u>spatial transcriptomics</u>, lets us see which genes are turned on in different parts of a tumour, while keeping their spatial positions. We applied this to several types of aggressive brain tumours, including <u>H3 G34-mutant</u> diffuse hemispheric gliomas (DHGs), pleomorphic xanthoastrocytomas (PXAs), embryonic tumours with multilayered rosettes (ETMRs), and atypical teratoid rhabdoid tumours (ATRTs).

We have generated matching <u>spatial proteomics</u> datasets for the same tumours.

#### **Current Focus**

Additional samples are being profiled using single-cell Methyl-HiC.

We designed our own custom 10X Xenium gene panels so we can study where specific genes are expressed within brain tumours. These panels include important genes related to the types of brain tumours we focus on, which aren't part of the standard Xenium panels. We will use them to analyze samples from high-grade gliomas.

For more details, please visit the ACCESS website or <u>first issue</u> of the newsletter.

#### **National Research Project:** Sarcoma

Project Name: Sarcoma MetAstasis Research • Taskforce, SMART Project

> Project Co-Leads: Livia Garzia, Rebecca Gladdy, and Poul Sorensen

#### **Brief Description**

In high-risk pediatric sarcomas, we have made progress in understanding the biology of the primary (original) tumours. However, we still do not fully understand what causes these cancers to relapse or spread to other parts of the body.  $\circ$ This project aims to explore gaps in knowledge and identify therapeutic targets that might be relevant to prevent or treat metastasis and relapse in pediatric high-risk sarcomas.

#### **Recent Achievements**

- All available metastatic and relapse sarcoma samples for the project have been prepared for sequencing, libraries passed quality controlled and are in queue for sequencing and potential proteomic analysis.
- o All patient-derived xenografts (PDXs) have been validated by bulk RNASeq collaboration with Adam Shlien. These PDXs are being prepared for spatial analysis.
- o Clinically relevant mouse models from the Gladdy lab have been profiled by multiome.

- o Plasma samples for the early detection of For more details, please visit the ACCESS metastasis and target selection have arrived in website or first issue of the newsletter. Vancouver for extracellular vesicle analysis, proteomics and RNASeg experiments.
- Members of the Garzia and Gladdy labs have received training on the Pulmonary Metastasis Assay (PuMA) from a staff scientist in the Sorensen lab. A tissue clearing protocol to image the lungs following the PuMA assay has been established and will be implemented as soon as validated PDX samples are available. To learn more about the PuMA assay, here is a link to an article from the Sorensen lab.
- We have made progress in developing novel approaches and tools for use in this project, including optimizing a protocol to evaluate the role of chondroitin sulfate proteoglycans on immune cell rewiring and pioneering a nanopore-based approach for the rapid diagnosis of fusion-positive sarcoma, as well as extending our knowledge of potential immunotherapy surface targets in Ewing sarcoma. osteosarcoma. and rhabdomyosarcoma tumours.

#### **Current Focus**

Our current focus is on plasma proteomics and RNASeq, sequencing of all metastatic and relapse samples, and spatial analysis of validated PDXs.



#### **National Research Platform: PCMM Network**

Project Name: Pediatric Cancer Models &

• Mechanisms (PCMM) Network

Project Lead: Chris Maxwell

#### **Brief Description**

The Pediatric Cancer Models and Mechanisms (PCMM) network is a national platform that has created an experts registry and a matching program for preclinical and clinical researchers. The PCMM network promotes preclinical investigations and enables all researchers across Canada to access leading experts and technologies.

#### **Achievements**

We have established the PCMM Network Experts Registry, which has registered 60+ investigators across the country and is connected to 1000+ domestic and international investigators. We have also launched two rounds of competition where 7 seed-funding projects have been supported for a total of \$400 000.

#### **Current Focus**

Research agreements are being finalized for projects approved from the second round.

We are encouraging investigators to take 10 minutes to register their expertise with the PCMM Network Experts Registry. This resource helps connect researchers in Canada and around the For more details, please visit the PCMM world with investigators with the expertise, technologies, and models, they seek.

Network Website and the PCMM Network Registry.

Q Search				Include resu	llts from partner registries
earch for res	earchers using at least one of the follow	ving criteria:			
Name					
Part of rese	earcher name				
Research inte	erests				
Keyword in	research description				
With at least	one of the following Tumor Tissue term	ns (or any if none are selecte	d):		
				0	
With at least	one of the following Research Technol	ogy terms (or any if none are	selected):		
☐ High-throu	ughput drug screening				
Cell therap	pies				
Proteomic	s				
Genomics					
Genome-e	diting				
Metabolon	nics				
☐ Epigenetic	s				
Bioinforma	atics				
☐ Flow cytor	netry/sorting				
Imaging (ii	n vivo, in vitro)				
Spatial pro	filing technologies				
With at least	one of the following Research Models	terms (or any if none are sele	ected):		
Patient-sp	ecific				
The same	use DDX - fish	☐ PDX - other	☐ Primary Organoid	☐ Primary Co-culture	
PDX - mou	pecific				
	c - Transgenic - fish	☐ Transgenic - other			
Disease-s					
Disease-sp Transgenic mouse					
Disease-sp Transgenic mouse	eathway-specific	☐ Frog	☐ Zebrafish	☐ Fly	□ Worm
Transgenic mouse Gene- or p Mouse Yeast	athway-specific			□ Fly	□ Worm

The PCMM Network Registry of Experts: The webpage (registry.pcmmnetwork.ca/sear ch) where investigators can search for investigators in Canada and around the world from the PCMM Network and partner registries. These searches can be filtered based on the name of the investigator, tumour tissue, research technologies, research models, genes, reactome pathways, research interests, and key words. You can also visit the website to register your expertise at the top right corner of the webpage.

# National Research Platform: Molecular Pathology

**Project Name:** Molecular Pathology Strategy

Project Co-Leads: Philipp Lange and Liana

Nobre



#### **Brief Description**

This project focuses on a comprehensive, research-driven strategy to identify and accelerate the integration of next-generation molecular pathology assays and platforms into routine clinical care, ensuring equitable access for all children with cancer across Canada. Our objectives include (i) establishing the necessary infrastructure to expand access to proteomic profiling; (ii) advancing the clinical implementation

of <u>liquid biopsy</u> technologies; and (iii) democratizing access to advanced molecular diagnostics and specialized expertise by creating a National Molecular Pathology Board (MPB) dedicated to pediatric oncology.

#### **Recent Achievements**

- Teams at each of the National Pediatric Proteomic Centres (NPPCs) that we are establishing have conducted deep proteome profiling of four personalized oncology cases — three at BC Children's Hospital (Lange) and one at SickKids (Moran). Two of the four cases originated from external institutions, helping us develop the infrastructure and pipelines to promote equitable access to proteome profiling across sites.
- The liquid biopsy team is launching a questionnaire to assess the landscape of available clinical and research assays performed on liquid biopsy samples with the goal of making these tests more accessible.
- The MPB has recently overcome an obstacle that delayed its activities. The Board reviews patient cases and connects them with advanced molecular assays that are otherwise difficult to access due to financial or logistical barriers. The Board has now resumed case reviews. For more information, please contact the Cancer Biology Theme Project Manager. Details and access to submission forms will soon be available on the MPB's dedicated

webpage on the ACCESS website, which is currently under development.

#### **Current Focus**

We are working on the official launch of the MPB with a dedicated webpage on the ACCESS website. Additionally, we are deploying a liquid biopsy questionnaire to assess the landscape of liquid biopsy assays, both clinical and research, relevant and available to the pediatric, adolescent, and young adult cancer patient cases in Canada. We are also finalizing contracts between participating institutions with available liquid biopsy assays.

In the coming days, we will be presenting the work of the NPPCs and the proteomics team at the Canadian Cancer Research Alliance Conference in Calgary and the Human Proteome Organization Conference in Toronto, both in November 2025.

#### **Requests & Opportunities**

We are looking for a second Person With Lived Experience to join the Liquid Biopsy project team.

For more details, please visit the ACCESS website or <u>first issue</u> of the newsletter.

# **Our Projects**

# National Research Platform: Modelling

Project Name: Pediatric Preclinical Modelling
Program

**Project Co-Leads:** Jason Berman, James Lim, and Donna Senger

#### **Brief Description**

Cancer treatments have often relied on strong chemotherapy drugs that harm both healthy and cancer cells and are chosen mainly by histologic cancer type instead of being personalized to each person's unique tumour. New genetic testing can personalize treatments, but results are still limited because they don't always show how a tumour actually responds to drugs prior to the patient receiving it. This project will create a national program that uses a patient's cancer cells to establish <u>patient-derived xenograft (PDX)</u> models to test how they react to different drugs, helping doctors choose the most effective, targeted treatments for young patients.

#### **Recent Achievements**

- Standard Operating Procedures are in place for <u>cryopreserving</u> patient samples and shipping fresh tissue. Material resources are also available to support institutes in properly cryopreserving patient samples for modelling.
- An intake form is available to enroll patients to the modelling program.

- o One test patient case has been modelled.
- Connections have been established with the <u>Infant Leukemia</u> and <u>SMART</u> (sarcoma) national research projects, as well as the Biobanking Network.
- We have expanded the modelling program with the addition of Dr. Jean-Philippe Babeu at the Centre Hospitalier Universitaire de Sherbrooke (CHUS) who works on organoid modelling.
- A recently published scientific article highlights the potential of zebrafish modelling. Modelling Program project co-lead, Jason Berman, and lab the CHEO at Research Institute/University of Ottawa, in collaboration with Precision Oncology For Young peopLE (PROFYLE) - a driver project within ACCESS - and Australia's Zero Childhood Cancer (ZERO) initiatives, have shown that preclinical zebrafish models can effectively support realtime clinical decision-making for difficult pediatric cancer cases. You can read the scientific article here, the ACCESS summary article here, and the CTV Ottawa news article here.

#### **Current Focus**

We are focused on obtaining research ethics approval to extend the modelling of cancers from patients beyond those enrolled in PROFYLE. We are also collaborating with the <u>Biobanking Network</u> on their virtual biobank and establishing

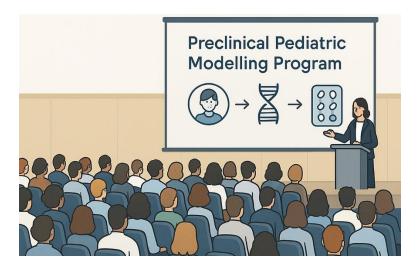
models and performing <u>drug screens</u> fo additional patient cases.

#### Requests & Opportunities

We are giving a talk on the Modelling Program at the Canadian Cancer Research Conference in Calgary (Nov 2-4, 2025).

We also continue to survey the Canadian pediatric cancer research community to identify investigators that have or are establishing PDX models and have the capacity or interest in evaluating drug responses to inform clinical implementation and improve patient outcomes.

For more details, please visit the ACCESS website and <u>first issue</u> of the newsletter.



# Events & Opportunities

### REQUESTS & OPPORTUNITIES

If you did not complete the **ACCESS Conflict of Interest** form from 2024, please do so by clicking on the link here.

If you are presenting or publishing work funded by ACCESS, please send abstracts and manuscripts to the ACCESS secretariat for review, well in advance of submission. For more information, you can access the **ACCESS Publication Policy** here.

If you have suggestions for stories, research highlights, collaborations, innovations, or milestones that would interest Cancer Biology Theme stakeholders, we'd love to hear from you. We also invite you to share your insights by contributing a short article—take a look at the "Featured Contributor" and "Featured Article" sections in this and past issues for inspiration.

The <u>Infant Leukemia</u> and <u>Liquid Biopsy</u> Project Teams are seeking **People With Lived Experience (PWLE)** interested in joining their teams. Please reach out to the Cancer Biology Theme Project Manager, if you are interested.

#### **EVENTS**

3<sup>rd</sup> Annual ACCESS General Meeting is taking place in Toronto from March 9<sup>th</sup>-11<sup>th</sup>, 2026 – mark your calendars!

#### IN CASE YOU MISSED IT

ACCESS 2024-2025 Impact Report \*NEW ACCESS Conflict of Interest Form ACCESS Publication Policy April2025

Cancer Biology Theme Newsletter Issue 1 (Jan 2025)

Cancer Biology Theme Newsletter: Issue 2 (Jun 2025)

PCMM Network Website PCMM Network Registry

FOR EVEN MORE INFORMATION, please reach out to the Cancer Biology Theme Project Manager, Emily Nakada, or ACCESS Project Manager, Tricia Schneider.

# Glossary

**Biomarkers:** biological molecules like genes and proteins that suggest the presence of cancer in a patient.

Chondroitin Sulfate Proteoglycans: large molecules found on the surface of cells and in the spaces between them. They can help tumour cells spread, avoid the immune system, or resist treatment, and are often studied as potential biomarkers or therapeutic targets in cancers.

Chromatin: a complex of DNA and proteins that form the chromosomes found in the cells. Its primary function is to package DNA so they are more compact.

**Circulating Tumour DNA (ctDNA):** DNA that is released from cancer cells into the bloodstream.

**Cryopreserving**: the process of freezing cells, tissues, or other biological materials so they can be safely stored and later thawed without being damaged.

**Driver Alteration:** a specific change in the sequence or expression level of a gene that provides a significant growth advantage to a cell, allowing it to proliferate abnormally.

**Drug Screen**: an experimental test used to identify compounds that produce a specific biological effect. Researchers expose cells, tissues, or purified proteins to a collection of

potential drugs—sometimes thousands at a time—to see which ones cause a desired response, such as killing tumour cells or blocking a disease-related pathway.

**Extracellular Vesicle (EV):** sacs released by cells into the space outside a cell but still within the respective tissue or organ.

Fusion Gene: an abnormal gene made when parts of two different genes combine, which can lead to the production of a new protein that may cause cancer or other diseases.

**Genomics**: the study of all DNA in a cell, tissue, or organ, to understand their structure, function, and interactions.

H3 G34R-Mutant: a genetic change affecting a histone protein (H3), which helps control how DNA is packaged and how genes are turned on or off. It is commonly found in pediatric high-grade gliomas and is linked to altered gene regulation and tumour development.

Heterogeneity: means diversity or differences within a group. In the context of tumour heterogeneity, it refers to how cells in a tumour that seem similar can still vary in important ways (i.e. at the molecular level – DNA, RNA, proteins).

Libraries: in the context of sequencing, libraries are created by breaking down extracted DNA or RNA into smaller fragments where special adapter sequences are added to the ends of these fragments. This is a necessary step to ensure samples are compatible with the sequencing machine, allowing it to be amplified and read accurately.

Liquid Biopsy: a minimally invasive laboratory test that analyzes bodily fluids to detect cancer cells or tumour DNA.

Longitudinal: something measured or observed over time, rather than just once. So, longitudinal testing follows the same patients or samples repeatedly, to see how things change or progress.

Methylation: a chemical change to DNA where small molecules called methyl groups are added to certain parts of the DNA sequence. It can turn genes on or off, affecting how cells behave. In cancer, the study of methylation profiles can help detect cancer or predict how it might respond to treatment.

Methyl-HiC: a technique that lets scientists study both the 3D structure of DNA and its chemical modifications (methylation).

Multiome: an approach that measures multiple types of molecular information from the same cell. For example, which genes are active and how the

DNA is organized — to give a more complete picture of how that cell works.

Nanopore: a method of analyzing molecules by passing them through tiny pores (nanopores) and measuring changes in electrical current to reveal the molecular structure or sequence in real time.

Next Generation Sequencing: technology that allows scientists to read and analyze the genetic material (DNA or RNA) of an organism quickly and in large amounts, providing detailed information about genes, mutations, and how they are expressed.

Patient-Derived Xenograft (PDX) Model: a model of cancer where a patient's tissue or cells are engrafted in an animal to accurately represent the biology and heterogeneity of a cancer.

Pharmacotyping: the process of analyzing an individual's/group's response to drugs based on their genetic, molecular, or cellular characteristics. It helps identify which treatments are likely to be most effective or safe, supporting more personalized approaches.

**Proteomics:** The study of the structures, composition, function, interactions and activities of proteins.

Pulmonary Metastasis Assay (PuMA): an experimental test that allows researchers to observe how cancer cells form and grow in lung

tissue, helping them understand the process of cancer spreading (metastasis).

RNASeg: the reading or sequencing of RNA to study which genes are active in a cell or tissue and how much they are expressed.

Spatial (Transcriptomics and Proteomics): methods that study the expression of genes and proteins across a tissue sample to understand how cells interact with each other and their environment.

Terms of Reference: a foundational document that defines the purpose, structure, and operating principles of a network or committee.

Transcriptome Sequencing: the technology that sequences and measures RNA to generate the data transcriptomics that researchers analyze.

**Transcriptomics:** the study of all RNA molecules in a cell, tissue, or organ at a specific time.

Whole Exome Sequencing (WES): a technique that reads the protein-coding parts of a person's DNA to find genetic changes that may cause disease.

Whole Genome Sequencing (**WGS**): a technique that decodes all of a person's DNA to identify genetic changes across the whole genome, not just in genes that code for proteins.



Thank you Merci



accessforkidscancer.ca





