



International User Group Meeting

22nd - 23rd April 2026 | Valencia

PROGRAMME





Jean-Pol Detiffe

OncoDNA founder

Dear Colleague,

By the time you read this you will already have arrived in Valencia, Spain for OncoDNA's 3rd International User Group. We aim for this meeting to be our 'biggest and best' yet!

Honestly, it's the highlight of my year seeing our customers and my teams sharing knowledge around our products, technology and services.

The event is designed to help build consensus on what best practice looks like, solve challenges, network, exchange tips and experiences with the overarching aim of supporting one another.

Whether you are providing input as a presenter, have submitted a poster or are simply participating as a delegate you are all as important to us because you are proactively contributing to our wider customer community.

This year I am delighted that Gareth Gerrard from Synnovis London is 'chairing' this meeting, evidencing our commitment to make the User Group ever more customer-led over time.

In celebration of your success we are pleased this year to be running customer awards and I look forward to presenting these awards during the meeting.

I would also like to extend a heartfelt thank you to the entire OncoDNA team. Their dedication, energy and attention to detail behind the scenes are what makes this event possible year after year, and truly brings this community to life.

No doubt the next two days will be fun, busy and tiring but Genomics is far easier when we work together!

All good wishes...

Jean-Pol Detiffe

- Your main contacts -

Meet your International User Group Meeting Support Team

To ensure a smooth and enjoyable experience throughout the User Group Meeting, a dedicated team is available to support you at every step.

Whether you have a question about the programme, need technical assistance, or encounter any organisational issue during the event, please don't hesitate to reach out to the contact persons listed below. They are here to help and make sure everything runs seamlessly for you.

We encourage you to get in touch at any time — your experience matters, and the team will be happy to assist.



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- Day 1 -

13:00 - 13:45	Delegate Lunch and Arrival
13:45 - 14:15	Welcome - Who is in the room and what do our customers want? Jean-Pol Detiffe , Founder OncoDNA and Chief Strategy and Innovation Office
14:15- 14:45	The customers role in driving innovation and how to work as a collaborative partner with OncoDNA Gareth Gerrard , Clinical Scientist and is the Scientific Director of the South East Genomic Laboratory Hub (SEGLH) / Synnovis Genomics
14:45 - 15:15	Collaborating for Growth - Why OncoDNA's Product Roadmap is focused on Customer Success Koenraad Eycken , Chief Product Officer, OncoDNA
15:15 - 16:00	Liquid Biopsy or Tissue NGS testing in Oncology - Competing Options or a Complementary Strategy? Petra Pekorna , Research Specialist, University Hospital Brno & Gerald Martin , Commercial Applications Specialist, OncoDNA Czech Republic, Belgium

Project/Research Breakouts

16:15 - 16:45	Room: Celeste Can DNA Biomarkers enhance Non-Muscle Invasive Bladder Cancer Surveillance? Evaluating the efficacy of Multimodal Genomic Profiling for bladder cancer recurrence and progression: A Prospective Longitudinal Study Amy Newman Deputy Operations Lead Cancer Genetics/Synnovis	Room: Bordeos Role of OncoDEEP in Resolving Histological Overlap: Exclusion of Secretory Breast Carcinoma in a Triple-Negative Tumour Rika Pienaar Oncologist/ Cancer Care	Room: Purpura Comprehensive Genomic Profiling (OncoDEEP) identifies potential therapeutic targets and reveals novel insights into the genomic landscape of extra skeletal myxoid chondrosarcoma Marcel Trautmann Gerhard-Domagk-Institute of Pathology / University Hospital Munster
17:00 - 17:30	Room: Celeste One Panel, One Pipeline: Implementing OncoDEEP (and OncoSELECT) for a streamlined routine diagnostic service Susanne Gonder Molecular Biologist, MVZ für HZMD Trier	Room: Bordeos Improving HRD detection through genomic instability signatures and pre-analytical DNA quality control Radoslava Rechterikova PHD Candidate, Oncology Institute of St. Elizabeth & Faculty of Medicine Comenius University	Room: Purpura Validation of the Large OncoDEEP RNA Panel: Our experience at the Hospital Clinic of Barcelona Eva Hernandez Illan Responsible Lead for Solid Tumour Diagnostics, Hospital Clinic of Barcelona
17:35 - 18:00	Reflections from Day One, Customer Awards* and Evening Plans *Speakers at the 2026 User Group are automatically entered into the customer awards - who will you choose to become a winner? Adriana Terradez , Chief Commercial Officer, OncoDNA		
18:30 - 20:00	A complimentary tour of Valencia to see the sites alongside your new found friends and colleagues (meet at the front reception)		
20:00 - 23:00	Dinner at the Restaurant "Contrapunto" (Queen Sofia Palace of Arts, Av. del Professor López Piñero, 1, Quatre Carreres, 46013 València)		

- Speakers on Day 1 -



Gareth Gerrard (Chair):

A Clinical Scientist and is the Scientific Director of the South East Genomic Laboratory Hub (SEGLH) / Synnovis Genomics (UK)

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Petra Pekorna:

Research Specialist, University Hospital Brno (CZ)

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Koenraad Eycken:

Chief Product Officer, OncoDNA (BE)

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Amy Newman:

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Gerald Martin:

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Marcel Trautmann:

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Rika Pienaar:

Oncologist / Cancer Care (ZA)

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Radoslava Rechterikova:

PHD Candidate, Oncology Institute of St. Elizabeth & Faculty of Medicine Comenius University (SL)

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Susanne Gonder:

Molecular Biologist, MVZ für HZMD Trier (GE)

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Eva Hernandez Illan,

Responsible Lead for Solid Tumour Diagnostics, Hospital Clinic of Barcelona (SP)

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Adriana Terradez

Chief Commercial Officer, OncoDNA (SP)

a.terradez@oncodna.com



- Day 2 -

07.45 - 08.45 **Morning walk:** Meet outside the front of the hotel with suitable footwear

09.00 - 09.15 **Award Winners and their tangible examples of innovation and influence**

09.15 - 10.00 **OncoXPLORE - Transitioning innovation to clinical routine.**

Gian Eeraerts,
PhD researcher, KU Leuven

Breakout sessions

10.15 - 10.45

Delegates choice

Room: Celeste

Integrating Longitudinal Genomic Profiling to Navigate Clonal Evolution and Multi-Pathway Resistance in Metastatic Breast Cancer

Cengiz Yakicier,
Medical Genetics, NPG Genetics Centre, Istanbul

Room: Bordeos

Validation of an automated OncoDEEP workflow using the Hamilton Star

Lucas Pavlou,
Clinical Scientist, Synnovis, London

10.45 - 11.15

Delegates choice

Room: Purpura

Insights from OncoDEEP validation on the Avitiz4 platform

Ioana Lemnian,
Head of Bioinformatics, PathoNext GMBH

Room: Bordeos

The experience of implementing OncoDEEP and OncoSELECT enabled by an MGI sequencer

Ceyda Centiner,
Oncology Department Manager, Geneks

11.15 - 11.45

Delegates choice

Room: Celeste

Navigating Validation Challenges at OncoDNA: From Reagents and Bioinformatics to Reporting in a Rapidly Evolving Environment and working towards CE-IVDR-Validated Solutions

Cedric Balsat,
Product Development Specialist at OncoDNA

Room: Bordeos

In-house analytical validation of HRD detection in HGSOC using the OncoDEEP assay for routine diagnostic implementation

Helene Blons,
Clinical Molecular Biologist, Hôpital Européen Georges Pompidou AP-HP, Paris

11.50 - 12.30

Customer Spotlight on challenging NGS Samples -A review of customers experiences (send in your samples to be considered as part of this presentation and discussion)

Sebastien Sauvage, Head of Scientific Support, OncoDNA

Networking and Lunch

13.15 - 14.00

Lifting the Lid on Reporting to shine a light onto the process and rules when generating reports using OncoKDM

Simon Lefevre, Production Manager, OncoDNA

14.00 - 14.45

Reporting 'live'. An interactive session uncovering what 'works well' and 'not so well' within delegates own laboratories. Solutions will be considered including how Chat GBT and AI are to be integrated in OncoKDM. The interactive session will conclude with the alignment of where reporting efficiencies can be found in laboratories and delegates drawing up a plan for how to move forward.

Facilitated by **Koenraad Eycken,** Chief Product Officer, OncoDNA & **Adriana Terradez,** Chief Commercial Officer, OncoDNA

14.45 - 15.15

Key Takeaways from User Group 2026 & User Group 2027 and how to get involved/Final Remarks from the Chair

Jean-Pol Detiffe, Founder OncoDNA and Chief Strategy and Innovation Officer, **Gareth Gerrard,** Clinical Scientist and is the Scientific Director of the South East Genomic Laboratory Hub (SEGLH) / Synnovis Genomics

- Speakers on Day 2 -



Gareth Gerrard (Chair):

A Clinical Scientist and is the Scientific Director of the South East Genomic Laboratory Hub (SEGLH) / Synnovis Genomics (UK)

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Chief Product Officer, OncoDNA (BE)

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- Presentation takeaways -

The customers role in driving innovation and how to work as a collaborative partner with OncoDNA

Gareth Gerrard,

A Clinical Scientist and is the Scientific Director of the South East Genomic Laboratory Hub (SEGLH) / Synnovis Genomics



Take Aways from your Chair



- | | |
|----|--|
| 01 | This is your User Group - what do you want to get out of it? |
| 02 | Some of you may want to lead and some may just want to listen – but all are participating by being here! |
| 03 | This is your opportunity to engage directly with other labs and to learn from each other's experience. |
| 04 | Different labs and different countries may have different ways of doing things – and different ways of engaging. |
| 05 | Who wants to take up the baton of chairing the next User Group meeting? |

Collaborating for Growth - Why OncoDNA's Product Roadmap is focused on Customer Success

Koenraad Eycken,

Chief Product Officer, OncoDNA



Take Aways



- ✓ OncoDNA is an oncology solutions provider with more than 12 years of experience in precision medicine that has transitioned from centralized to decentralized testing
- ✓ OncoDNA products, features and functionalities have evolved and will require further evolution based on your customer feedback, market needs and implementation of new technologies
- ✓ The Product portfolio is being expanded into liquid biopsies as well as adding new functionality into the OncoKDM platform
- ✓ OncoDNA teams across Sale, Scientific, Support and Bioinformatics aim to stay close to our customers through various forums



- Presentation takeaways -

Liquid Biopsy or Tissue NGS testing in Oncology - Competing Options or a Complementary Strategy?



Petra Pekorna, Research Specialist, University Hospital Brno & **Gerald Martin**, Commercial Applications Specialist, OncoDNA Czech Republic, Belgium

Take Aways



- ✓ Take care to formulate a testing strategy that draws upon both solid and liquid testing
- ✓ Create enough capacity to test in the laboratory to respond to demand from the clinic in a timely manner
- ✓ Draft a plan which includes your testing targets (goals, diseases and genes)
- ✓ Construct your validation plan and document the outcomes required
- ✓ Consider changes in context overtime (clinical requirements, assays, automation, staffing and logistics)
- ✓ Document routes to reimbursement as they are evolving all the time!

Can DNA Biomarkers enhance Non-Muscle Invasive Bladder Cancer Surveillance? Evaluating the efficacy of Multimodal Genomic Profiling for bladder cancer recurrence and progression: A Prospective Longitudinal Study



Amy Newman
Deputy Operations Lead Cancer Genetics/Synnovis

Take Aways



01	✓ DNA extracted from urine can be sequenced successfully using the OncoDEEP assay, producing excellent quality metrics and high concordance with standard of care
02	✓ Preliminary studies in a small cohort suggests OncoDEEP can detect the presence of tumour in new diagnosis, recurrence and progression of non-muscle invasive bladder cancer
03	✓ More low-grade non-muscle invasive urine samples are needed for robust statistical analysis
04	✓ Strategic planning, regular engagement and 'buy-in' from clinicians, regulatory bodies, patients and other stakeholders is essential to deliver timely and successful research projects



- Presentation takeaways -

Role of OncoDEEP in Resolving Histological Overlap: Exclusion of Secretory Breast Carcinoma in a Triple-Negative Tumour

Rika Pienaar
Oncologist/ Cancer Care



Take Aways



01	Definitive Diagnosis: While traditional methods like histology and immunohistochemistry (IHC) can suggest the subtype, only NGS can definitively identify the ETV6-NTRK3 fusion in SBC cases, which is crucial for distinguishing it from other TNBCs.
02	Avoiding overtreatment: Identifying the ETV6-NTRK3 fusion confirms a diagnosis of SBC, preventing the potential overtreatment of a patient who might otherwise be categorized under the more aggressive 'basal-like TNBC' umbrella
03	Guiding Therapy in Advanced Disease: Identifying the NTRK gene fusion is crucial because it is a tumour-agnostic biomarker that predicts a response to specific targeted therapies, such as NTRK inhibitors (e.g., larotrectinib and entrectinib). This information helps oncologists plan precise treatment strategies.

Makretsov N, He M, Hayes M, et al. A fluorescence in situ hybridization study of ETV6-NTRK3 fusion gene in secretory breast carcinoma. *Genes Chromosomes Cancer* 2004;40:152-157.

Comprehensive Genomic Profiling (OncoDEEP) identifies potential therapeutic targets and reveals novel insights into the genomic landscape of extra skeletal myxoid chondrosarcoma

Marcel Trautmann
Gerhard-Domagk-Institute of Pathology /University Hospital Munster



Take Aways



- ✓ **Extraskeletal myxoid chondrosarcomas (EMC)** are rare mesenchymal neoplasms, accompanied by high rates of recurrence and metastases.
- ✓ The molecular hallmarks of EMCs are cytogenetic *NR4A3* rearrangements, generating chimeric **NR4A3 fusion oncoproteins**.
- ✓ **Genomic landscape of EMC – Comprehensive genomic profile / OncoDEEP:**
>1,700 variants have been identified in 6g tissue specimens:
48% of pts with novel pathogenic or likely pathogenic variants
TMB high (1.4%) & HRD pos. (2.9%) pts



Novel therapeutic actionability with potential clinical benefits identified by CGP via in-house use of the OncoDEEP kit



- Presentation takeaways -

One Panel, One Pipeline: Implementing OncoDEEP (and OncoSELECT) for a streamlined routine diagnostic service

Susanne Gonder*
Molecular Biologist, MVZ für HZMD Trier



Take Aways

- 01 ✓ Implementation of OncoDEEP is a multifactorial process and it does not begin in the laboratory...
- 02 ✓ DNA and RNA Quality is a critical factor in molecular pathology
- 03 ✓ The customer is king and continuous improvement is essential



Improving HRD detection through genomic instability signatures and pre-analytical DNA quality control

Radoslava Rechterikova
PHD Candidate, Oncology Institute of St. Elizabeth & Faculty of Medicine Comenius University



Take Aways

- ✓ HRD is not limited to BRCA1/2 → genomic scar-based testing improves detection
- ✓ BRCA-only testing misses clinically relevant patients
- ✓ HRD+ and HRD- tumours represent distinct biological subtypes
- ✓ HRR gene mutations ≠ functional HRD → genomic context matters
- ✓ DNA integrity (DV900), not concentration, determines data reliability
- ✓ NGS may underestimate MSI → orthogonal validation is recommended



Main message: Integrated HRD assessment + DNA quality control are essential for accurate, clinically actionable results



- Presentation takeaways -

Validation of the Large OncoDEEP RNA Panel: Our experience at the Hospital Clinic of Barcelona



Eva Hernandez Illan
Responsible Lead for Solid Tumour Diagnostics, Hospital Clinic of Barcelona

Take Aways



- High performance for RNA fusions detection.**
More informative than most current panels (especially **amplicon-based** panels), with a higher likelihood of identifying **new or uncommon fusion partners**.
Robust performance in challenging samples, including low-input material and cytological specimens.
 - Longer turnaround time** (compared to amplicon-based panels).
Batch-dependent workflow, (compared to amplicon-based panels).
Challenging interpretation of fusion results, need to establish or improve understanding of fusion-calling thresholds
Room for improvement in data visualization and reporting within OncoKDM: including better display of fusions and imbalances and RNA quality metrics; unified RNA result tables in the website.
- ↳ Feedback provided working to inform development of OncoKDM

OncoXPLORE - Transitioning innovation to clinical routine



Gian Eraerts:
PhD researcher, KU Leuven

Take Aways



- | | |
|----|---|
| 01 | OncoXPLORE is able to detect actionable alterations with high sensitivity in clinical use cases with unmet diagnostic needs |
| 02 | Cost-effective, scalable and fast (<7 TAT) off-the shelf solution |
| 03 | (Epi)genomics improves cancer signal detection and profiling |
| 04 | Need for further clinical validation and collaboration to increase knowledge about (epi)genomic profiles in multiple tumor types |



- Presentation takeaways -

Integrating Longitudinal Genomic Profiling to Navigate Clonal Evolution and Multi-Pathway Resistance in Metastatic Breast Cancer



Cengiz Yakicier,
Medical Genetics, NPG Genetics Centre, Istanbul

Take Aways



01	<ul style="list-style-type: none">✓ Continuous Genomic Surveillance is Imperative<ul style="list-style-type: none">✓ Proactive Therapy Switching<ul style="list-style-type: none">✓ <i>Don't wait for systematic clinical progression</i>✓ Design Combination Strategies<ul style="list-style-type: none">✓ <i>Cut off evolutionary routes</i>✓ Precision Trial Matching<ul style="list-style-type: none">✓ Real time biology dictates the trial not the baseline history
02	✓ Repeated, Genomic Assessment is Essential to Outpace Complex, Adaptive Resistance
03	<p>Supporting Conclusion:</p> <p>A static, single-point-in-time biopsy is insufficient for metastatic breast cancer, Only continuous profiling enables the multi-pronged targeting of both the HER2 kinase domain and the P13K pathway necessary to manage dynamic resistance</p>

Insights from OncoDEEP validation on the Aviti24 platform

Ioana Lemnian,
Head of Bioinformatics, PathoNext GMBH



Take Aways



01	✓ OncoDEEP performs robustly on AVITI24 and the synergy between sequencing platform and assay has reduced our hands-on time in the wetlab
02	✓ We are reaching higher coverages and a better uniformity of the coverage with less sequencing data per sample.
03	✓ OncoDEEP on AVITI24 is a flexible and cost-efficient solution for CGP (we were able to reduce our costs by ~ 35%).
04	✓ Longer sequencing runtime and dry lab part lead currently to longer turn-around times, but we look forward to finetuning our workflow, especially having the constant support from OncoDNA.



- Presentation takeaways -

The experience of implementing OncoDEEP and OncoSELECT enabled by an MGI sequencer



Ceyda Centiner,
Oncology Department Manager, Genoks

Take Aways



- ✓ The platform-independent wet-lab workflow of the **OncoDEEP and OncoSELECT kits**, with identical protocols, facilitated a streamlined validation exercise
- ✓ The open manipulation capability on T7 sequencing allowed rapid validation and optimization, facilitated by our wet-lab expertise.
- ✓ Mixed-run capability allows simultaneous processing of different materials, regardless of index / adaptors used.
- ✓ A flexible, open, and robust workflow enabling rapid validation, consistent throughput, and scalable sequencing performance on **MGI T7** instrument.

Navigating Validation Challenges at OncoDNA: From Reagents and Bioinformatics to Reporting in a Rapidly Evolving Environment and working towards CE-IVDR-Validated Solutions



Cedric Balsat,
Product Development Specialist at OncoDNA

Take Aways



01	Rapid and continuous evolving environment
02	Validation must be continuous, not static integrating the cutting edge of innovation
03	End to end validation is essential
04	Evolving regulatory context (IVDR)- Increases regulatory requirements and documentation
05	A risk-based analytical approach to ensure OncoDNA solutions continue to evolves whilst complying with IVDR regulations



- Presentation takeaways -

In-house analytical validation of HRD detection in HGSOC using the OncoDEEP assay for routine diagnostic implementation

Helene Blons,
Clinical Molecular Biologist, Hôpital Européen Georges Pompidou AP-HP, Paris



Take Aways



1. The OncoDEEP kit offers a all in one solution for HRR gene mutation testing and HRD scoring
2. It was clinically validated on PAOLA samples
3. The HRD score shows decreased sensitivity in samples with TP53 VAF <30%, a limitation that has also been reported with alternative HRD scoring approaches
4. Particular caution is advised when interpreting results, as single-exon deletions and pathogenic splice-region mutations may be challenging to detect

- ✓ Use enriched macrodissected samples if possible
- ✓ Use TP53 VAF to validate HRD score « if BRCAwt »
- ✓ Check the VCF file or the « all variant section » for splice region variants
- ✓ Check the CNV section for large deletions in HRR genes
- ✓ Communicate with OncoDNA staff when needed

Customer Spotlight on challenging NGS Samples -A review of customers experiences (send in your samples to be considered as part of this presentation and discussion)

Sebastien Sauvage,
Head of Scientific Support, OncoDNA



Take Aways



Remember Next Generation Sequencing, Comprehensive Genomic Profiling (CGP) is tricky because its not just about Sequencing!

- It's actually a multi-dimensional optimisation problem:
- Noisy Biological Input + Complex Variant Types + Imperfect Computational Models + High Clinical stakes
- And add in the 'Human Dimension' which introduces additional operational variability and risk:

An NGS sample can be challenging because Clinical CGP workflows are strongly influenced by variation in:

- Tissue Biopsy Collection within the Operating Theatre Environment
- Biopsy Tissue Storage and Transport Conditions
- Environmental Conditions in the Laboratory
- Multiple Manual Steps (Wet Lab, QC Checks, Curation)
- Interpretation Between and Amongst Experts
- Differences in Laboratory Practices, Pipelines And Reporting Standards

👉 **All the above introduces variability, potential errors, and reproducibility challenges when analysing NGS samples**



- Presentation takeaways -

Lifting the Lid on Reporting to shine a light onto the process and rules when generating reports using OncoKDM

Simon Lefevre,
Production Manager, OncoDNA



Take Aways



- 01 ✓ OncoKDM was initially developed to deliver actionable NGS results for Oncologists
- 02 ✓ OncoDNA moving to a decentralized model has launched a significant transformation of OncoKDM
- 03 ✓ OncoKDM is now being further developed to meet the needs of our laboratory customers
- 04 ✓ OncoKDM is your platform so work with us to ensure it adds value to your laboratory reporting workflows

Validation of an automated OncoDEEP workflow using the Hamilton Star

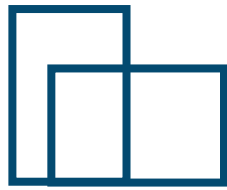
Lucas Pavlou,
Clinical Scientist, Synnovis, London



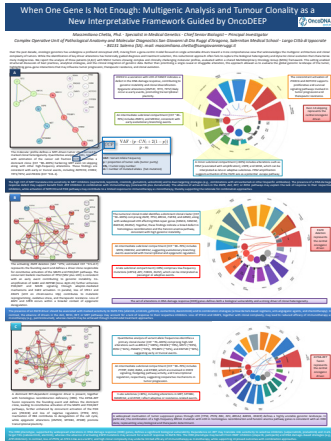
Take Aways



- 01 Tight deadlines, collaboration and flexible planning are required
- 02 Challenges around troubleshooting and ongoing/further development post implementation
- 03 Automation – different problems not no problems
- 04 Priorities during development should be simplifying changes, implementing robustness and rigorous testing for error potential and optimisation
- 05 Validation of an automated OncoDEEP workflow has led to improved testing capacity and reduced TAT



- Posters -

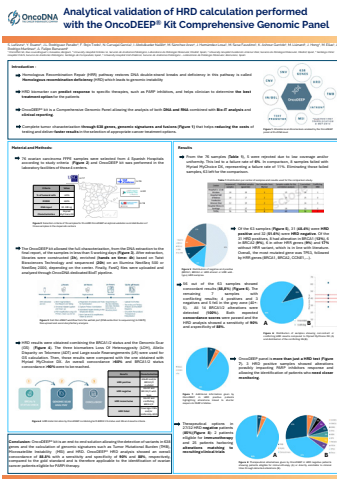


When One Gene Is Not Enough: Multigenic Analysis and Tumour Clonality as a New Interpretative Framework Guided by OncoDEEP

This poster explores how over the past decade, oncologic genomics has undergone a profound conceptual shift, moving from a gene-centric model focused on single actionable drivers toward a more comprehensive view that acknowledges the multigenic architecture and clonal complexity of tumors. While the identification of key driver alterations has historically guided diagnosis and treatment selection, this reductionist approach often fails to capture the biological heterogeneity and dynamic clonal evolution that characterize many malignancies.

In this study, we report the analysis of eight patients with tumors showing complex and clinically challenging molecular profiles, evaluated within a shared Multidisciplinary Oncology Group (MOG) framework. This collaborative setting enabled structured discussion of best practices, analytical strategies, and the clinical integration of genomic data into more nuanced, dynamic, and personalized management of cancer patients.

Massimiliano Chetta, Principal Investigator, UOC Anatomia Patologica



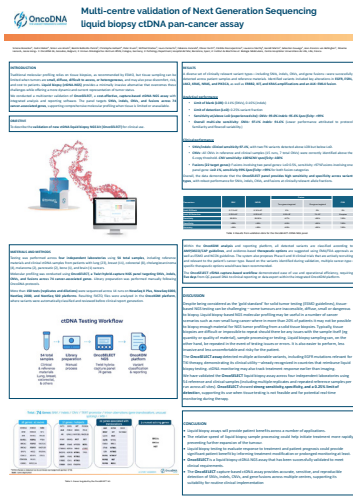
Analytical validation of HRD calculation performed with the OncoDEEP® Kit Comprehensive Genomic Panel

This poster explores the clinical relevance of homologous recombination deficiency (HRD) and its integration into comprehensive genomic profiling for improved treatment decision-making. The Homologous Recombination Repair (HRR) pathway plays a critical role in restoring DNA double-strand breaks. Deficiency in this pathway, referred to as homologous recombination deficiency (HRD), leads to genomic instability and contributes to tumour progression.

As a biomarker, HRD enables the prediction of response to targeted therapies such as PARP inhibitors, supporting clinicians in selecting the most appropriate treatment options for patients.

The OncoDEEP® kit is a comprehensive genomic panel that enables the combined analysis of DNA and RNA, supported by advanced Bio-IT analysis and clinical reporting. By covering 638 genes, genomic signatures, and gene fusions (Figure 1), it allows for complete tumour characterisation, reduces the need for multiple sequential tests, and accelerates access to appropriate cancer treatment options.

S. Lefèvre, Y. Ruano, J.L. Rodriguez Peralto, F. Rojo Todo, N. Carvajal Garcia, I. Abdulkader Nallib, M. Sánchez Ares, J. Hernández Losa, M. Sese Faustino, K. Ashour Garrido, M. Lienard, J. Hong, M. Elias, A.B. Rodrigo Martinez, A. Felipe Benavent



Multi-centre validation of Next Generation Sequencing liquid biopsy ctDNA pan-cancer assay

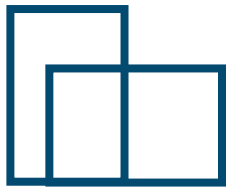
This poster explores the limitations of traditional tissue-based molecular profiling and evaluates the potential of liquid biopsy as a complementary or alternative approach.

Traditional molecular profiling relies on tissue biopsies, as recommended by the European Society for Medical Oncology. However, tissue sampling can be challenging when tumours are small, diffuse, difficult to access, or highly heterogeneous. In addition, biopsy procedures may involve patient discomfort, procedural risks, and increased costs.

Liquid biopsy based on circulating tumour DNA (ctDNA-NGS) offers a minimally invasive alternative that helps overcome these limitations, while providing a more dynamic and real-time representation of tumour status.

In this study, we conducted a multicentre validation of OncoSELECT, a cost-effective, capture-based ctDNA NGS assay with integrated analysis and reporting software. The panel targets single nucleotide variants (SNVs), insertions/deletions (indels), copy number variations (CNVs), and gene fusions across 74 cancer-associated genes, enabling comprehensive molecular profiling when tissue samples are limited or unavailable.

Simone Bonecker1, Cedric Balsat, Simon von Arnell, Beatriz Bellosillo Paricio, Christophe Hallaert, Peter Krusin, Wilfried Stücker, Laura Camacho, Fabienne Escande, Olivier Farchi, Clotilde Descarpentries, Laurence Stechly, Gerald Martin, Sebastien Sauvage, Jean-Francois van Bellinghen, Maxime Lienard, Jessie Hong.



- Posters -

OncoDEEP Revolution: Comprehensive Somatic Variant Analysis in Solid Tumors – Our Experiences

Faculty of Life Sciences, Department of Pathology and Molecular Medicine, Second Faculty of Medicine, Charles University and Masaryk Memorial Cancer Institute, Prague, Czech Republic

INTRODUCTION
OncoDEEP is a comprehensive genomic profiling (CGP) platform designed to analyze both known and heretofore undiscovered variants using a combination of DNA and RNA sequencing. In our laboratory, OncoDEEP is primarily applied to detect potential driver mutations for identifying potential therapeutic options in solid tumors. However, its use extends to clinical research, including the identification of novel biomarkers and the discovery of potential drug targets.

METHODS
217 tumor samples (102 solid tumors and 115 pediatric samples) collected from 2024-2025. The samples represent 102 different tumor types. OncoDEEP is a comprehensive genomic profiling (CGP) platform designed to analyze both known and heretofore undiscovered variants using a combination of DNA and RNA sequencing. In our laboratory, OncoDEEP is primarily applied to detect potential driver mutations for identifying potential therapeutic options in solid tumors. However, its use extends to clinical research, including the identification of novel biomarkers and the discovery of potential drug targets.

RESULTS
OncoDEEP identified 10,000 somatic variants across 217 tumor samples. The most frequent variants were found in the EGFR, KRAS, and BRAF genes. The results were compared with other NGS platforms, showing high concordance.

CONCLUSIONS
OncoDEEP is a highly suitable assay for comprehensive genomic profiling (CGP) of solid tumors, providing an all-in-one solution for clinical research and patient care. The platform's ability to detect both known and novel variants makes it a valuable tool for identifying potential therapeutic options and drug targets.

OncoDEEP Revolution: Comprehensive Somatic Variant Analysis in Solid Tumors – Our Experiences

This poster explores how our laboratory has validated and generated high confidence in results using the OncoDEEP kit with high sensitivity and specificity. Using this kit, we support clinical applications in precision oncology by identifying clinically relevant somatic alterations (SNV, insertions, deletions, CNV, RNA fusions) and comprehensive genetic biomarkers such as HRD, MSI, and TMB, across a spectrum of solid tumors. A main advantage of the OncoDEEP kit is its easy integration, more efficient work-flow, rapid turnaround time, and use of the OncoKDM® platform for interpretation of the NGS data.

Using the OncoDEEP/OncoKDM combination we are able to identify predictive or diagnostic biomarkers for each patient in a single test. The final report presents detected variants with clinical significance associated with potential therapeutic impact (or lack of) according to FDA/EMA/NCCN/ESMO guidelines. The average turnaround time from sample receipt to release of the final report (PDF and printed copy sent to the referring clinician) is approximately 15 – 20 days.

Jana Čopíková, Clinical Laboratory Geneticist, Motol and Homolka University Hospital

Implementation of OncoDEEP® in Routine Diagnostics: A Case Study of Fibrous Hamartoma of Infancy

Dr. Marina Wierz, Dr. Zuzana Šimková, Dr. Heide Arent, Molekulardiagnostische Tumor Genetik, Prof. Dr. med. Christoph Lohmann-Vass, Pathologie Trier, Germany

1. Background
Fibrous hamartoma of infancy (FHI) is a rare, benign, fibrous tumor of the skin, typically occurring in the first year of life. It is characterized by a proliferation of fibroblasts and adipocytes, often associated with a hamartomatous component.

2. Clinical Presentation
The patient presented with a large, soft, fleshy, and painless mass on the lower leg, which had been present since birth. The mass was well-circumscribed and showed no signs of malignancy.

3. Diagnostics
Histopathological examination of the tumor revealed a characteristic triphasic pattern consisting of spindle-shaped fibroblasts, adipocytes, and a hamartomatous component. Immunohistochemical staining for CD34 and SMA was performed.

4.1 Histopathology
Microscopic images showing the characteristic triphasic pattern of FHI, including spindle-shaped fibroblasts, adipocytes, and a hamartomatous component.

4.2 Molecular Analysis
Genomic profiling using OncoDEEP revealed a duplication in exon 20 of the EGFR gene, supporting the diagnosis of Fibrous Hamartoma of Infancy.

5. Discussion & Conclusions
This case highlights the importance of molecular analysis in the diagnosis of rare entities. The identification of a duplication in exon 20 of the EGFR gene provides strong evidence for the diagnosis of FHI.

6. Acknowledgments
We thank the OncoDEEP team for their support and expertise in the molecular analysis of this case.

Implementation of OncoDEEP® in Routine Diagnostics: A Case Study of Fibrous Hamartoma of Infancy

This poster addresses Subcutaneous Spindle Cell Lesions in Infants which are diagnostically challenging due to overlapping clinical and histopathological features. We report a 7-month-old infant with multiple newly developed cutaneous and subcutaneous lesions and profuse, "salty" sweating. To clarify the diagnosis, molecular analysis was performed using the OncoDEEP kit. Genetic testing revealed a duplication in exon 20 of the EGFR gene, supporting the diagnosis of Fibrous Hamartoma of Infancy. This case underscores the value of integrating routine molecular diagnostics into paediatric dermatopathology to enable accurate classification of rare entities.

Marina Wierz, Molecular Biologist MVZ für HZMD Trier

A NOVEL SOMATIC BRAF EXON 3 SKIPPING MUTATION IN LEIOMYOSARCOMA: A CASE REPORT

MSc. EKIN KÖNİ, DR. MURAT BOYUKODAN, DR. ARDA KENLİ, DR. N. CENGİZ YAKİCİR

BACKGROUND
Leiomyosarcoma (LMS) is a rare and aggressive mesenchymal malignancy with limited therapeutic options in advanced stages. Comprehensive genomic profiling (CGP) enables identification of clinically actionable and therapeutic targets.

BIOLOGICAL MECHANISM
BRAF Exon 3 encodes part of the BRAF-binding domain (BRD) and plays a role in the MAPK pathway. Exon 3 skipping leads to loss of autoinhibitory regulation, resulting in constitutive activation of the MAPK pathway (BRAF-MEK-ERK).

CASE PRESENTATION
Patient diagnosed with Leiomyosarcoma. Tumor content: 50%. Analysis performed using OncoDEEP. 48-gene CGP panel. Report date: 30 December 2025.

TREATMENT STRATEGY
Non-V600E BRAF alterations may not respond to classical BRAF inhibitors. MEK inhibition (e.g., Trametinib) represents a rational therapeutic strategy. KRAS/NRAS wild-type status supports BRAF as the primary oncogenic driver.

DISCUSSION
This case highlights a rare non-V600E BRAF alteration in Leiomyosarcoma. CGP is essential to identify clinically actionable variants that may be missed by standard diagnostics. The detected alteration provides a strong rationale for targeted therapy via MEK pathway inhibition. These findings emphasize the role of precision oncology in rare and aggressive malignancies.

RESULTS
Genomic profiling revealed a novel somatic BRAF Exon 3 skipping mutation. Additional somatic alterations identified include TP53, V600E, NRAS, PIK3CA, SETD, and PTEN. LMS-associated genes also identified include CDKN2A, RB1, PTEN, ATRX, CDKN2A, and CDKN1A. Additional variants (high VAF) include MCL1 and NOTCH4.

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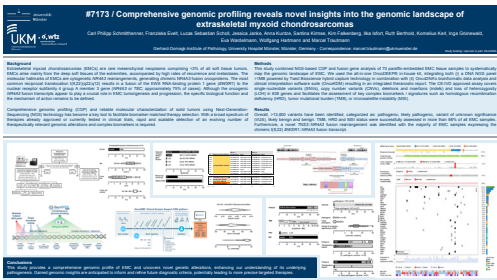
A Novel Somatic BRAF Exon 3 Skipping Mutation in Leiomyosarcoma: A Case Report

This poster addresses Aggressive Malignancy: Leiomyosarcoma (LMS) a rare and aggressive mesenchymal malignancy with limited therapeutic options in advanced stages. In the case presented Comprehensive Genomic Profiling (CGP) was performed using the OncoDNA 638-gene panel. The analysis identified a main driver BRAF Exon 3 skipping mutation and KEAP1 single-copy loss leading to the development of an Off-Label Treatment Strategy underpinned by a clear rationale. This case underscores the necessity of CGP in Leiomyosarcoma to identify rare, targetable mutations that standard diagnostics might miss.

Cengiz Yakicier, Medial Genetics MPG Genetic Centre

Comprehensive genomic profiling reveals novel insights into the genomic landscape of extraskeletal myxoid chondrosarcomas

This poster explores the genomic landscape of extraskeletal myxoid chondrosarcomas (EMCs) and highlights the value of comprehensive genomic profiling for improved molecular characterisation and therapeutic insights.



Extraskeletal myxoid chondrosarcomas (EMCs) are rare mesenchymal neoplasms, accounting for less than 3% of all soft tissue tumours. They typically arise in the deep soft tissues of the extremities and are associated with high rates of recurrence and metastasis. At the molecular level, EMCs are characterised by NR4A3 gene rearrangements, leading to the formation of oncogenic fusion proteins. The most common translocation, t(9;22)(q22;q12), results in an EWSR1::NR4A3 fusion, observed in approximately 75% of cases. While these fusion transcripts are believed to play a central role in tumour development, their precise biological function and mechanisms of action remain to be fully elucidated.

Comprehensive genomic profiling (CGP) using Next-Generation Sequencing (NGS) has become a key approach to enable biomarker-driven therapy selection. With an increasing number of targeted therapies available or under clinical investigation, there is a growing need for rapid and scalable detection of clinically relevant genomic alterations and complex biomarkers.

Carl Philipp Schmittthener, Franziska Evelt, Lucas Sebastian Scholl, Jessica Janke, Anna Kuntze, Santina Kirmse, Kim Falkenberg, Ilka Isfort, Ruth Berthold, Kornelius Kerl, Inga Grünewald, Eva Wardelmann, Wolfgang Hartmann and Marcel Trautmann

Clinical validation on the PAOLA-1/ENGOT-ov25 cohort of HRD calculation performed with the OncoDEEP® Kit comprehensive genomic panel.

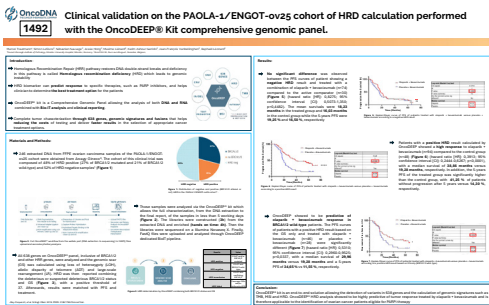
This poster explores the role of homologous recombination deficiency (HRD) as a key biomarker in cancer and its integration into comprehensive genomic profiling strategies.

The Homologous Recombination Repair (HRR) pathway is responsible for repairing DNA double-strand breaks. Deficiency in this pathway, known as homologous recombination deficiency (HRD), leads to genomic instability and contributes to tumour development and progression.

HRD has emerged as a clinically relevant biomarker, enabling the prediction of response to targeted therapies such as PARP inhibitors, and supporting clinicians in selecting the most appropriate treatment strategies for patients.

The OncoDEEP® kit is a comprehensive genomic profiling solution that enables the combined analysis of DNA and RNA, supported by advanced Bio-IT analysis and clinical reporting. By covering 638 genes, genomic signatures, and gene fusions, it provides a complete characterisation of tumours while helping to reduce the need for multiple sequential tests and associated costs.

Marcel Trautmann, Simon Lefèvre, Sébastien Sauvage, Jessie Hong, Maxime Liénard, Karim Ashour Garrido, Jean-François Vanbellinghen, Raphaël Léonard



- Your Field Application Support Team -



Jean-Francois Vanbellinghen



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Our Field Application Specialist (FAS) customer support team is here to assist you throughout the meeting and beyond. For any technical questions, operational support or product-related inquiries, feel free to reach out at customersupport@oncodna.com

. The team will be happy to support you and ensure you get the most out of your OncoDNA experience.

- Partners -



At Twist Bioscience, we work in service of customers who are changing the world for the better. In fields such as medicine, agriculture and industrial chemicals, by using our synthetic DNA tools, our customers are developing ways to better lives and improve the sustainability of the planet. The faster our customers succeed, the better for all of us, and Twist Bioscience is uniquely positioned to help accelerate their efforts.

Our innovative silicon-based DNA Synthesis Platform provides precision at a scale that is otherwise unavailable to our customers. Our platform technologies overcome inefficiencies and enable cost-effective, rapid, precise, high-throughput synthesis, sequencing and therapeutics discovery, providing both the quality and quantity of the tools they need to most rapidly realize the opportunity ahead. For more information about our products and services, please visit www.twistbioscience.com.

- About OncoDNA -

Founded in 2012 OncoDNA has grown exponentially since its creation, closing milestone partnerships with leading oncology organisations and expanding its footprint worldwide. As a result, it has become a leading genomic and theranostic company translating molecular science and digital knowledge into clinical expertise to improve cancer treatment. OncoDNA's continued mission is to optimise cancer patients' treatment journeys by empowering health professionals, companies, and researchers to deliver the promise of precision medicine. OncoDNA offers a unique portfolio that combines NGS services, biomarker testing, data analysis software and clinical decision support tools. The OncoDNA Group is headquartered in Belgium, and its entities, BioSequence and IntegraGen are based in Spain and France. OncoDNA employs over 100 employees across multiple countries and works with an international network of Distributors to extend the reach of its work.

