

Why patients want biomarkers

Perspectives from a Cancer of Unknown Primary patient advocate
On a mission to #MakeTheMysteryHistory

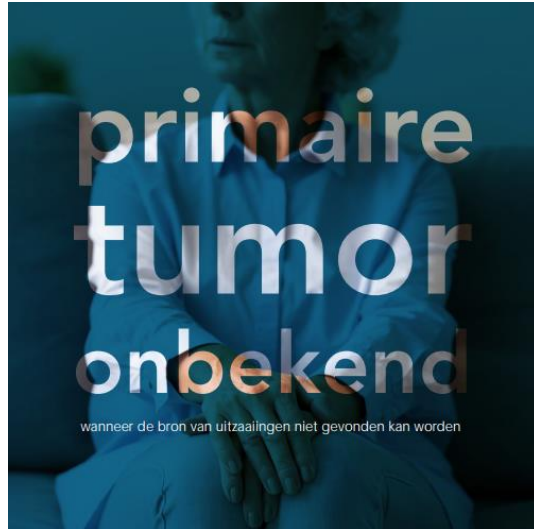
CDDF Workshop – November 2023



Why Missie Tumor Onbekend? Hederik's story



Missie Tumor Onbekend: Dutch CUP patient & advocacy organisation



- Circa **30 new diagnoses per week / 1,500 per year** in the Netherlands cancer registry
- **4th most common metastatic cancer** (together with pancreas), **2-5% of cancers across the globe**
- Patients did not fit well in existing care provision, they undergo **on average more diagnostics than other cancer patients**, but without result & the diagnostics trajectory does not finish
- **2 out of 3 CUP patients did not get a treatment** including palliative treatment
 - In contrast to patients with metastases from known primary tumors, who only in 1 out of 3 cases did not get a treatment
- **>50% of patients died within 2 months after their first hospital visit**
- **1-year survival rate of 17%, long-term (5-year survival) of only 5%**

Our goals

1. Raise awareness, and get the 'unknown' out of CUP
2. Increase quality of care & life for patients and their families that deal with CUP with treatment and support
3. Increase the chance of survival & decrease CUP incidence

Our ultimate ambition: No more newly-diagnosed patients, a perspective towards a cure & the best quality of care

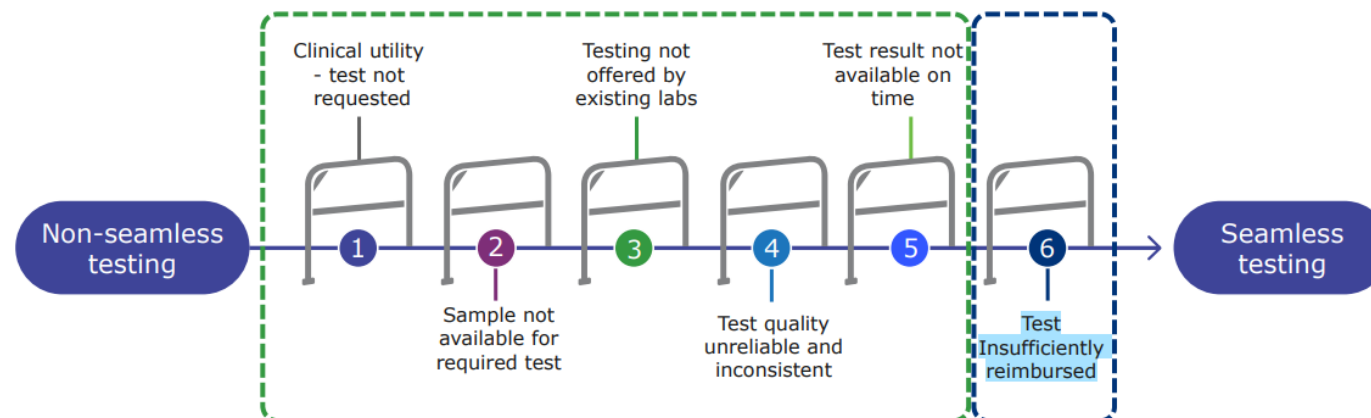
Starting in 2020, raising awareness and overcoming barriers was key



More **awareness and information about CUP** was necessary among patients/relatives, medical specialists & nurses, the broader healthcare sector, politics, and the general public

Could **biomarker testing/advanced molecular diagnostics** be of help **to overcome the diagnostics bottle neck** in CUP?

Several **barriers towards implementation** existed at the time (2020/21):



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Feasibility of whole-genome sequencing-based tumor diagnostics in routine pathology practice

Kris G Samsom, Luuk J Schipper, Paul Roepman, Linda JW Bosch, Ferry Lalezari, Elisabeth G Klompshouwer, Adrianus J de Langen, Tineke E Buffart, Immy Riethorst, Lieke Schoenmaker, Daojin Schout, Vincent van der Noort, Jose G van den Berg, Ewart de Bruijn, Jacobus JM van der Hoeven, Hans van Snellenberg, Lizet E van der Kolk, Edwin Cuppen, Emile E Voest, Gerrit A Meijer , Kim Monkhorst ... [See fewer authors](#) ^

Our 2021 campaign supported access to biomarker testing

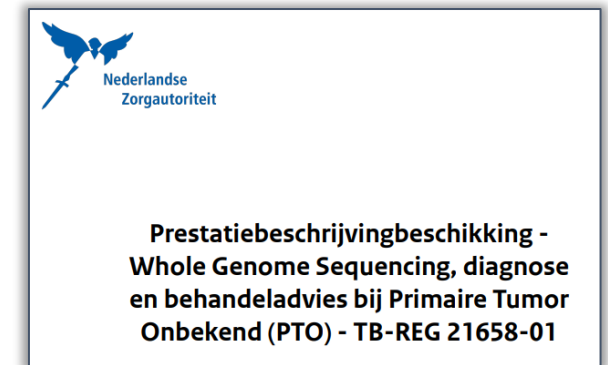


Successful lobby with medical specialists, health insurers, politicians and national health authority for **reimbursement from the national basic healthcare insurance package for WGS for patients with (suspected) CUP**

Since April 2021 when WGS became reimbursed care for CUP:

- **8 CUP outpatient clinics/expertise teams have emerged in centers with an MTB**
- **A national diagnostic care pathway** has been launched in collaboration with the oncologist (NVMO) and pathologist (NVVP) scientific associations
- The **2012 national clinical guideline for CUP** is in the process of getting updated

We have a **proactive collaboration with (inter)national experts on new scientific studies, care improvements, access to information & patient support both in the Netherlands as well as internationally**



Promising first results of access to biomarker testing in Dutch CUP patients

- **Timely use of biomarker testing works¹:**
 - In c. **68%** of patients **WGS is of value-add in identifying the primary tumor**
 - Actionable events with **matched therapy options in clinical trials were identified in 47% of patients**
 - Hence, a **treatment consequence/recommendation** could be found in c. **80% of CUP patients** that receive **WGS**
 - In clinical practice we see that c. **70% of patients start on a molecularly guided therapy**
 - Most patients can **find access to treatment** due to the innovative **DRUP-trial** protocol
- Other **positive “side effects”**:
 - Increase in CUP patients being discussed in MTBs → **increase in expertise building**
 - **Patients see expert clinicians sooner and diagnostic work-up could be completed sooner, hence also (palliative) support could be provided sooner**
 - Mistaken CUP cases referred to CUP clinics can be solved sooner
 - Despite WGS being reimbursed, **other biomarker tests are provided to more CUP patients** to help solve the case
 - **Local collaboration** between CUP clinics and regional referral hospitals is enhanced
- The **urgency** is there: we need to test patients timely so patients can be treated timely as well



Complete genomic characterization in patients with cancer of unknown primary origin in routine diagnostics
 L.J. Schipper^{1,8*}, K.G. Samsom^{2*}, P. Snaebjornsson², T. Battaglia⁷, L.J.W. Bosch², F. Lalezari³, P. Priestley⁴, C. Shale⁴, A.J. van den Broek⁵, N. Jacobs⁵, P. Roepman⁵, J.J.M. van der Hoeven⁵, N. Steeghs⁶, M.A. Vollebergh⁶, S. Marchetti⁷, E. Cuppen^{5,8,9}, G.A. Meijer², E.E. Voest^{1,6,9}, K. Monkhorst^{2†}

Biomarker testing should become available in the ‘toolkit’ of a medical specialist in a center of expertise within a ‘learning care system’

¹ Schipper LJ, Samsom KG, Snaebjornsson P, Battaglia T, Bosch LJW, Lalezari F, Priestley P, Shale C, van den Broek AJ, Jacobs N, Roepman P, van der Hoeven JJM, Steeghs N, Vollebergh MA, Marchetti S, Cuppen E, Meijer GA, Voest EE, Monkhorst K. Complete genomic characterization in patients with cancer of unknown primary origin in routine diagnostics. ESMO Open. 2022 Dec;7(6):100611. doi: 10.1016/j.esmoop.2022.100611. Epub 2022 Dec 1. PMID: 36463731; PMCID: PMC9808446.

A patient perspective on a 'learning care system' for biomarker testing

1. **A large majority of EU-based cancer patients are willing to share their medical data for re-use for scientific & clinical purposes**
 - A small majority **prefer to automatically share** their data
2. In a 'learning care system' data collection is the standard and occurs in an almost natural, cohesive, secure and pragmatic way:
 - **Investments in data infrastructure** at the country, regional and hospital level are a priority to start the right way
 - Informed consent: a patient should be able to **easily opt-in/out to share their data**, and this should occur **from the start of the patient journey**
 - **PROMs and quality of life** data are also collected and help in validating the value patients have for the care received
 - Future research or input for clinical standards could be derived from the real-world data
 - **Knowledge sharing** should go both out and in of the 'learning care system'
 - Governance: an **independent third party** to manage the data collection could help
3. **The 'learning care system' should lead to quicker recognition for health innovations from health authorities and reimbursement agencies**
 - **Are registration and reimbursement frameworks tailored yet for a more agnostic approach?**
 - **Can treatments registered for a first indication be expanded to other indications and patient (sub)-populations based on the data?**
4. We should better understand **at what point in the patient journey biomarker testing is most effective**, and how to then organize that for patients
5. The existing strict separation between care provision and research is challenging
 - **(Near) real-time data from actual clinical setting are required**, and not only temporarily or later in a research setting

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