

Tracking glioma in cerebrospinal fluid, draw by draw.

Malignant glioma · tumour-informed ctDNA · nine CSF draws across four months, with a plasma baseline

An adult patient with malignant glioma needed close molecular follow-up. A baseline plasma sample carried almost no signal, consistent with the limited tumour DNA that central nervous system tumours shed into blood. Monitoring then moved to cerebrospinal fluid (CSF), where the same patient-specific assay tracked an abundant, changing signal across nine draws, including through a treatment intervention.

THE BRIEF

Epistamai Bio asked Simsen to move quickly: design a bespoke assay from the patient's existing tumour and germline sequencing, then test plasma and CSF to see whether tumour signal was detectable. If it was, the plan was to monitor at regular intervals, several times a month.

WHAT WE DID

From the patient's own whole exome and matched normal data, we built a personalised SiMSen-Seq panel of 16 patient-specific somatic variants, validated 27 days after receiving the tumour profile. A baseline plasma sample was tested first, then we monitored CSF across nine timepoints from February to June 2026, reporting per-variant and aggregate ctDNA load each time.

27 days

From tumour profile to validated, bespoke panel

16

Patient-specific variants tracked per sample

10

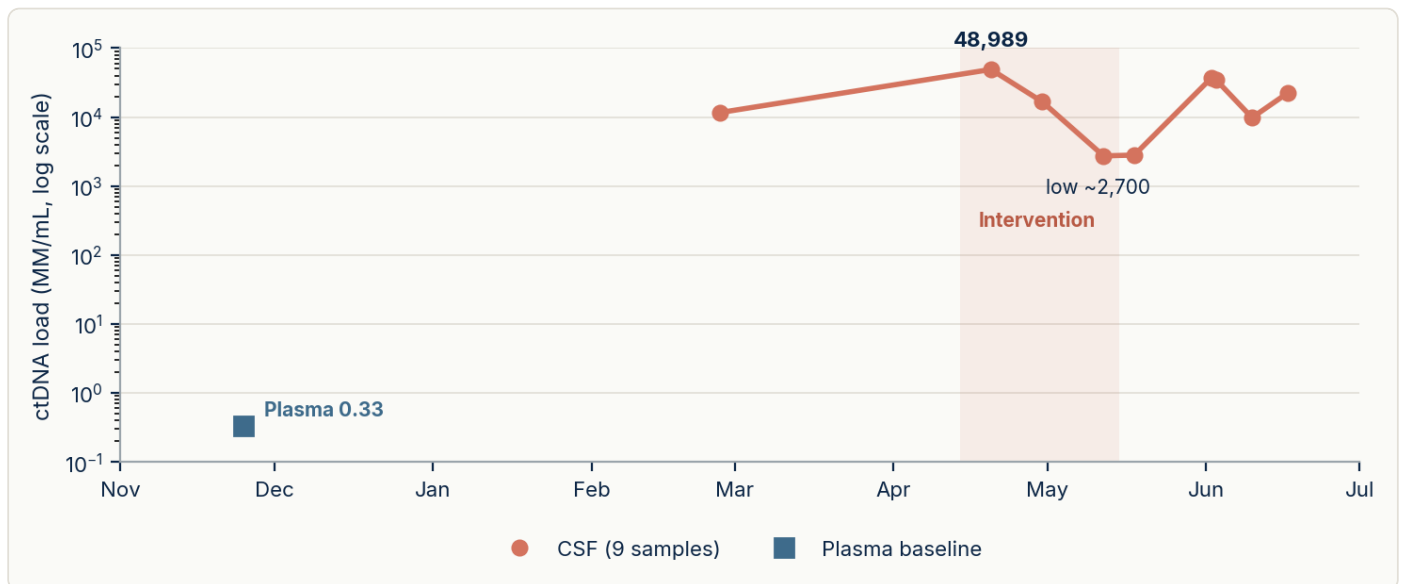
Timepoints: one plasma baseline, nine CSF

0.33 MM/mL

Baseline plasma load, just above the positivity threshold

2,700 to 49,000 MM/mL

Range across the nine CSF samples



Aggregate ctDNA load (mutant molecules per mL, log scale) at each timepoint. The CSF series (terracotta) carried abundant signal throughout, with an April peak, a mid-May low around the time of a treatment intervention, and a June rebound. The single plasma baseline (steel blue), taken months earlier, sat just above the positivity threshold.

WHAT WE FOUND

The baseline plasma sample showed an aggregate ctDNA load of 0.33 mutant molecules per mL, just above the positivity threshold. In CSF, the assay measured far more: between roughly 2,700 and 49,000 mutant molecules per mL across the nine samples. The signal tracked the treatment course, rising to an April peak, falling to a mid-May low around the time of a treatment intervention, and rebounding through June. One draw returned 11,592 MM/mL from just 1.5 ng of input cell-free DNA, a level of signal the high tumour fraction in CSF makes possible. Reading those dynamics, including how the molecular trend sits alongside stable interval imaging, is for the clinical team and tumour board; what the data show is that CSF gave a sensitive, repeatable readout at the cadence Epistamai Bio wanted.

"Treatment response monitoring in glioma remains highly limited, particularly in patients with diffuse or leptomeningeal disease where imaging can be difficult to interpret and where therapeutic decisions often need to be made before clear radiographic changes are available. The hope is that a CSF-based personalised ctDNA monitoring approach can provide a more dynamic readout of tumour burden and clonal evolution, enabling novel therapies to be tested and adapted more rationally over time."

Imran Chaudhry, MD · Epistamai Bio

CLINICAL TRIALS

For CNS indications, where a plasma sample may carry little tumour DNA, CSF can provide a measurable, longitudinal readout from one tumour-informed panel, supporting correlative endpoints and adaptive decisions.

CANCER CLINICS

The same bespoke assay runs on the sample type that carries the signal. For brain tumours that means CSF, tracked as often as the clinical question requires.

RESEARCH

Per-variant and aggregate data with an open, auditable pipeline, built from the patient's own tumour profile, suited to method work and co-publication.

Method and standing. SiMSen-Seq chemistry: PCR-based molecular barcoding with unique molecular identifiers (UMIs) and consensus error suppression to single-molecule level (Stahlberg et al., Nature Protocols, 2017; Nucleic Acids Research, 2016). CSF can reflect the genomic profile of central nervous system tumours more fully than plasma (De Mattos-Arruda et al., Nature Communications, 2015). The assay is used at VHIO, Erasmus MC and Sahlgrenska University Hospital, among other European centres, and Simsen is an assay provider in the GUIDE.MRD consortium on ctDNA standardisation. Samples stay within the EU under GDPR.

PANEL

16 patient-specific variants here; typically 20 to 30, tumour-informed. Actionable or resistance markers can be added on request.

SAMPLE

CSF and plasma both supported on the same panel. Matched normal needed once: existing germline data, or an EDTA tube at baseline.

SENSITIVITY

Single-variant detection ~0.01% VAF; aggregate sensitivity ~0.001% (10 ppm) across the tracked variants.

TURNAROUND

7 working days fast track; 12 standard, from sample receipt.

REPORTING

Per-variant MM/mL and VAF, aggregated ctDNA load, longitudinal timeline.

DELIVERY

Full-service EU lab, or SiMSen-Seq in your lab via LabSuite.

For research use only. Simsen Personal is not a diagnostic test and does not determine treatment. A positive result does not indicate a diagnosis of cancer; a negative result does not indicate remission. Results are specific to the assessed timepoint. The test has not been cleared by the FDA, EMA or other regulatory bodies. Patient details have been removed; this case study is published with Epistamai Bio's consent.

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