Serious incident report into the unexpected deterioration of a young person on the Acute Medical Unit SI619

This report contains the following attachments:

Att A and A1 The original serious incident report and action plan

Three reports from external experts

Att B1 Report from Dr Jon Durnian Consultant Ophthalmologist. A summary is included in the main paper.

Att B2 Report from Dr Shawn Halpin Consultant Neuroradiologist. A summary is included in the main paper.

Att B3 Report from Dr Michael Gross Consultant Neurologist. A summary is included in the main paper.

Att B3.1 Correspondence between Dr G and the patient's mother further to the report (six letters). A summary is included in the main paper.

Att C Investigation timeline

1 Introduction

The serious incident report into the death of a 25-year-old was completed in December 2021.

Due to concerns raised by the patient's mother the trust agreed in July 2022 to seeking independent review of the patient's care by a neurologist.

In August 2022 the patient's mother requested external reviews from a neuro-ophthalmologist, a neuro-radiologist and a neuro-intensivist as well as a neurologist. The trust agreed and sent proposed terms of reference to the patient's mother in November 2022. These were approved by the patient's mother in November 2023 who also supplied a list of documents and records that the experts were to consider, and the process of obtaining external experts was initiated. It was agreed that the patient's mother would select the external experts.

Reports have been received from the consultant ophthalmologist, consultant neuro radiologist and consultant neurologist. It was agreed with the patient's mother that each expert would receive the report from the previous expert, so these reports were requested sequentially.

A decision was made in December 2025 by the Trust to complete the serious incident investigation process and not to pursue the further report from a neuro-intensivist. The patient's mother was advised that this was due to our concern about two public demonstrations she and her associates undertook on Harley Street and subsequently outside the Royal College of Radiologists focusing on the radiologist who had reported on the first scan. The Trust's concern was that instead of seeking to learn from the external reports, these reports were being used to target an individual clinician. This was not the mutually agreed purpose of the reviews and is contrary to the collaborative way in which we had been hoping to work.

This report includes the original serious incident report and action plan and the external reviews from the three experts.

It also includes a summary of the issues that the patient's mother has raised in various correspondence. A timeline of the incident investigation is attached

2.Background

2.1 Serious incident report updated (post inquest) February 2022

The patient was a previously healthy 25-year-old young person who became acutely unwell with headache and vomiting, followed by altered mental state and unusual behaviour. The patient was admitted to ED on 17th July at 11pm. On initial assessment the patient was also noted to be moderately hyponatraemic (to have serum sodium below 130 mmol/L). The next day the patient had an emergency CT brain scan (initially reported as showing no acute intracranial finding), treatment for possible meningoencephalitis, and attempted lumbar puncture. This was unsuccessful as the patient was agitated and reported headache and the procedure was abandoned. During a second attempted procedure the patient suffered a respiratory arrest. By this time the patient had also become profoundly hyponatraemic. The patient was rapidly resuscitated, but a second CT scan showed generalised brain swelling with low lying cerebellar tonsils. The patient was admitted to the Intensive Care Unit on 19th July but tragically brain stem death was confirmed three days later on 22nd July 2021.

Care Delivery Problems (CDP)

CDP 1: Lack of monitoring of the patient's serum sodium, administration of three litres of intravenous fluid without relevant investigations having been performed, and the response to profound hyponatraemia on the afternoon of 18 July 2021

CDP 2: An emergency CT head scan not arranged on admission as it should have been.

These CDP were then subject to root cause analysis which led to the recommendations.

Recommendations

1. Guidance on the care of patients with hyponatraemia to be reviewed at local Clinical Governance meetings (Acute Medical Unit and Emergency Department).
2. Creation of a guideline on hyponatraemia for the Trust's Medical Emergency Document Library.
3. Ensure formal teaching on the care of patients with neurological presentations - and in particular patients with altered mental state / behaviour - is included in the rolling training programmes in Acute Medicine and Emergency Medicine.
4. To ensure formal regular teaching on hyponatraemia is included in the rolling training programmes in Acute Medicine and Emergency Medicine.
5. Review of the patient's initial CT scan at the Imaging Department's Learning Meeting.

Lessons Learned (learning which did not contribute to the incident however could be improved)

1. Importance of trying to contact family for background information to inform the care of patients unable to give a coherent account themselves (particularly in a time of pandemic).
2. Requirement for documented regular neurological observations for a patient with altered mental state.

2.2 Action plan

This is attached

2.3 Inquest

The inquest was held on 14th Feb 2022 and was a narrative determination as follows:

[redacted] **- determination on 14.02.22**

This has been an inquest on behalf of Our Sovereign Lady the Queen by me, Mary Elizabeth Hassell, Senior Coroner for Inner North London, touching the death of the Honourable [redacted] who died on 21 July 2021 at University College Hospital, 235 Euston Road, London.

I make a narrative determination as follows.

"The patient died from cerebral oedema. The cause of this remains unclear.
It is possible that the cause of the cerebral oedema was hyponatraemia. If the cause was hyponatremia, more monitoring and better clinical management would have afforded the patient a better chance of survival. A CT scan was not conducted as it should have been immediately following their admission to hospital on 17 July 2021. It is unclear what it would have shown if it had been conducted. A CT scan conducted the following day showed subtle signs of raised intracranial pressure, but this was not diagnosed at the time. If an earlier CT had been conducted and had shown raised intracranial pressure, or if the later scan had been reported as showing raised intracranial pressure, this would have changed the clinical management. A lumbar puncture would not have been attempted, although in any event the two lumbar punctures attempted did not puncture the dura and so did not impact on the outcome. However, knowing of the intracranial pressure would have resulted in head up nursing, transfer to intensive care and potential intubation. All of this would have afforded the patient a better chance of survival."

That concludes this inquest.

3 Summary of main concerns from the mother

3.1 Fundoscopy / ophthalmoscopy (examination of the back of the eye) was not successfully undertaken.

The Trust's serious incident (SI) report drew attention to the importance of ophthalmoscopy both on initial assessment and before lumbar puncture, if that were possible; and noted that abnormalities may be seen in patients with raised intracranial pressure. A UCLH consultant neurologist has advised that if it had been possible to examine the fundi, it is possible clinicians would have seen early papilloedema (indicating raised intracranial pressure).

A patient needs to be calm and still to perform a fundoscopy which was understandably not possible for the patient given how disorientated they were at times. In such circumstances, we would rely on a CT scan and this would have been an equivalent diagnostic approach in this case ie it was therefore necessary to rely on CT scan to determine whether there was any finding - such as evidence of brain swelling - which would be a contraindication to lumbar puncture.

On request from the patient's mother the Trust has provided details of training in ophthalmoscopy at undergraduate and postgraduate level and information on the availability of ophthalmoscopes.

3.2 Hyperammonaemia

Testing for high blood ammonia levels (hyperammonaemia) was not done and the PM describes this as a missed opportunity, linked to concern about a possible missed diagnosis of an inherited metabolic disease and hyperammonaemia.

The Trust has acknowledged that this was not considered in the Trust's serious incident investigation because none of the clinicians from the various specialties concerned considered hyperammonaemia as a possibility.

Reliable results for a blood ammonia level could not be obtained post-mortem. But other relevant biochemical tests were performed; and genetic testing was followed up – see section on genetic testing.

The patient was a previously well adult who experienced very rapid onset, acute cerebral oedema (swelling of the brain) with resulting encephalopathy (abnormal functioning of the brain). The patient's blood gas results normalised rather than worsened over time. Accordingly, the priority at the time was considered to be investigation for a vascular, drug or infective cause; it is possible that had the patient not deteriorated so quickly, ammonia testing would have been considered once other, more typical causes had been excluded.

The external neuroradiologist includes in his report hyperammonaemia as a possible cause for the findings on the patient's scans. But he notes the absence of abnormality in the liver biopsy and the negative genetic screen; and that characteristically hyperammonaemia is associated with sparing of some areas of the brain, although with more extreme cases the whole of the cerebrum might be affected.

It is agreed a blood ammonia level should be measured in all patients with encephalopathy without obvious alternative cause (intoxication, infection, trauma or vascular) / in whom the obvious causes have been excluded.

The Trust has now included ammonia in the acute confusional state guideline in the Medical Emergency Document Library. This also links to related conditions including hyponatraemia.

Testing for an ammonia level was introduced in the update to the Royal College of Emergency Medicine's Best Practice Guideline on Acute Behavioural Disturbance in Emergency Departments (Version 2 October 2023).

3.3 Genetic testing

After the patient's inquest, the coroner received an email from an endocrine specialist who read news coverage and suggested porphyria as a possible cause.

The coroner forwarded this to Professor Sheaff (pathologist), who did not think porphyria was supported by the autopsy findings but suggested possible genetic testing, as some metabolic diseases can only be detected this way.

UCLH consulted a metabolic diseases specialist at UCLH, who agreed porphyria was unlikely but proposed ornithine transcarbamylase (OTC) deficiency, a rare X-linked urea cycle disorder causing hyperammonaemia, as a plausible cause of the patient's encephalopathy. That is to say hyperammonaemia might have been a cause of the patient's presentation and OTC deficiency would have been the most likely cause of hyperammonaemia.

Genetic testing for OTC deficiency was agreed with the patient's mother in August 2022. Dr Robin Lachmann (RL) Consultant in Inherited Metabolic Disease agreed to meet with the patient's mother to explain genetic testing options.

RL contacted NHS Blood and Transplant (NHSBT) who manage amongst other areas organ transplantation, to ask for their help and discuss testing options. NHSBT contacted the hospital who are looking after the patient who received the patient's liver to request recipient tissue testing. The patient's mother was put into direct contact with NHSBT.

The request was for sequencing analysis of genes associated with hyperammonaemia on DNA extracted from a biopsy of the donated liver to rule out a diagnosis of ornithine carbamoyltransferase (OTC) deficiency in the donor.

Genetic testing was undertaken on a sample of DNA extracted from the donor liver (ie the patient's DNA) which showed no evidence of an underlying urea cycle disorder (including OTC deficiency). The liver recipient has also had biochemical testing done which also shows no evidence of urea cycle dysfunction. This is effectively testing the patient's liver and urea cycle and shows no evidence of an underlying urea cycle disorder. This is a critical piece of evidence.

In May 2023 the results of the liver recipient were shared with the PM. The report summarised that a genetic cause for the patient's disorder had not been identified. The report noted that 42 genes were screened.

The results make it less likely that the donor had an underlying urea cycle disorder. RL confirmed that genetic testing is not 100% sensitive, particularly for OTC deficiency and there are some patients with biochemical OTC deficiency in whom a genetic cause cannot be found. There are also other inherited metabolic diseases which cause a secondary hyperammonaemia, and metabolic causes of encephalopathy which are not associated with hyperammonaemia. However there are other biochemical and genetic tests which could be done to further explore whether the donor had an undiagnosed inherited metabolic disorder and further genetic testing for other disorders which can present with encephalopathy

The patient's mother confirmed that she would like to explore this possibility.

In July 2023 RL followed up with NHSBT regarding sample availability and tests to be undertaken and arranged for testing. Agreement to proceed was confirmed by the PM in November 2023.

Biochemical results from samples collected from the patient post mortem were available on 21st May 2024. As they were post mortem they were not straightforward to interpret but showed no evidence of hyperammonaemia.

Some genetic tests were available on 7th February 2025. The tests did not find any genetic changes to suggest the patient had an underlying inherited metabolic disease.

In August 2025 a further genetic report noted that the entire mitochondrial genome from the spleen sample has been sequenced. This has been reported as showing 'no clearly pathogenic mtDNA variant' and 'no genetic cause for the clinical presentation'

In summary there was extensive genetic testing on a sample of the patient's DNA which looks at over 750 genes associated with inborn errors of metabolism. This covers the urea cycle disorders (including OTC deficiency) and also many other possible causes of hyperammonaemia. No clinically relevant genetic changes were identified.

3.4 Hyponatraemia

The patient's mother has expressed concern about the place given to hyponatraemia in the Trust's serious incident investigation report; considers hyponatraemia to have been secondary to raised intracranial pressure due to another primary cause; and has drawn attention to a published series in which patients who died with hyponatraemia died from an underlying condition associated with hyponatraemia rather than from their low sodium.

This is a summary of a complex area in which the Trust's responses have been based on detailed advice by one of our consultant endocrinologists.

The cause of the patient's hyponatraemia is uncertain because the result of an early urinary sodium or urine and plasma osmolalities was not available– omissions for which the Trust has accepted and apologised. Possibilities include inappropriate secretion of antidiuretic hormone (IADH), although no primary cause for this has been identified on imaging or at post-mortem; and overhydration following salt loss from sweating on a hot day (as is seen in people who exercise).

The Trust's serious incident report was unable to say to what extent the patient's hyponatraemia was a contributory cause, or effect, of encephalopathy in the absence of more complete information.

Uncertainty about the cause and significance of the patient's hyponatraemia notwithstanding, the Trust has acknowledged that this feature of their illness should have been investigated and treated in its own right. Early investigations of serum and urine osmolality and urine sodium, in conjunction with clinical assessment, would have helped elucidate the cause of hyponatraemia and inform treatment.

The Trust has apologised for the shortcomings in the treatment of the patient's hyponatraemia.

A guideline on hyponatraemia has been added to the Trust's Medical Emergency Document Library, to support clinicians treating patients with hyponatraemia.

3.5 Initial CT scan and attempted lumbar puncture

The serious incident investigation noted there was a spectrum of views on whether the first scan showed generalised brain swelling. A consultant neurologist involved in the care of the patient considered the scan showed brain swelling. Subsequent review by two general radiologists for the serious incident report concluded "*changes on the initial CT were very subtle and of uncertain significance. We only became suspicious once the patient had deteriorated. A repeat CT at this stage was still only subtly abnormal but the abnormalities had progressed slightly.*"

The external neuroradiologist's review reports gross global cerebral and cerebellar swelling which is in contrast to the serious incident report findings.

Once the consultant neurologist had seen what he considered brain swelling on the CT scan, clinicians moved as quickly as possible to stop the second attempt at lumbar puncture; but by the time the SpR. contacted the Acute Medicine junior doctor, the patient had already deteriorated.

The serious incident investigation found that it seemed unlikely (though not impossible) that the patient's deterioration was due to the attempted lumbar puncture: no cerebrospinal fluid was obtained on attempted lumbar puncture, and the junior doctors reported they did not get a 'flashback' of any kind [when aspirating to confirm the position of the needle tip before injecting local anaesthetic].

4 Summary of external reports

4.1 Consultant ophthalmologist April 2025

Mr Jonathan Durnian (JD), a consultant ophthalmologist with expertise in neuro-ophthalmology, reviewed the earlier eye imaging and medical records to determine whether there had been any signs of raised intracranial pressure before the patient's final illness. He found that all optical coherence tomography (OCT) scans from 2019—both from Specsavers and Moorfields—were completely normal. There was no optic disc swelling, and the patient's slightly tilted optic discs were simply a common anatomical variation. At Moorfields, the patient's vision and eye examination were fully normal, and the presence of spontaneous venous pulsations showed that her intracranial pressure at that time was normal.

Because there was no papilloedema (optic disc swelling due to raised intracranial pressure) no abnormal OCT findings, and spontaneous venous pulsation were recorded as being seen on both eyes, JD concludes that there was no evidence of idiopathic intracranial hypertension or any malignant variant when the patient was seen in 2019. Nothing in the patient's earlier ophthalmic assessment suggested that they were at risk of the severe cerebral oedema that occurred in 2021.

4.2 Consultant neuroradiologist

Dr Shawn Halpin (SH) reviewed the brain imaging from July 2021 and concluded that the patient's first CT scan, taken early on 18 July is very abnormal. He describes clear evidence of widespread brain swelling, compression of the ventricles and basal cisterns, and both upward and downward herniation of the cerebellum. The scan was incorrectly reported as normal at the time. SH states that, had the findings been recognised, a lumbar puncture would not have been attempted, as the degree of hindbrain herniation made the procedure absolutely contraindicated.

The patient's condition continued to deteriorate, and later scans that day showed worsening swelling and more pronounced herniation. A CT angiogram that evening revealed an absence of blood flow in the internal carotid and intracranial arteries, a pattern Dr SH says is incompatible with life. MRI imaging on 20 July confirmed devastating, global and irreversible brain injury with widespread loss of blood perfusion and extensive damage to both the cerebral hemispheres and the cerebellum. In his view, the patient's injury was almost certainly unsurvivable from the evening of 18 July.

SH concludes that the patient died from a massive cerebral insult resulting in global cerebral and cerebellar perfusion failure. Although he considers a toxic or metabolic cause the most plausible category, he cannot identify a specific underlying condition. He notes that the most likely intrinsic metabolic origin would be within the urea cycle/ammonia pathway, although he notes the absence of abnormality in the biopsy taken from the patient's liver and the negative genetic screen. He wonders if it is possible that the patient unknowingly ingested a neurotoxic substance.

4.3 Consultant neurologist

Dr Michael Gross (MG) submitted a report and entered into further correspondence with the patient's mother.

4.3.1 Summary of report

MG reviewed the events surrounding the patient's rapid deterioration and death in July 2021. He describes how the patient became suddenly unwell with a severe headache, vomiting and confusion, and how their behaviour deteriorated throughout the night while remaining unaccompanied due to COVID restrictions. He is critical of the repeated assumption that the patient might be intoxicated, which he believes distracted clinicians from recognising a serious neurological illness.

He considers that by the early hours of 18 July, the patient's symptoms should have prompted urgent scanning and immediate treatment for suspected encephalitis or encephalopathy. He notes that the first CT scan, incorrectly reported as normal, actually showed severe brain swelling and early herniation, and he believes this error contributed to the decision to attempt a lumbar puncture, which was contraindicated, given that finding. Although he does not think the {attempted} lumbar puncture directly caused the patient's collapse, he concludes that the catastrophic brain oedema was already far advanced and ultimately unsurvivable.

Overall, MG identifies delays in recognising raised intracranial pressure, delays in senior review and treatment, and the impact of pandemic-driven barriers to proper history-taking. He acknowledges that although the initial assessments were poor and scanning might have been done a little earlier, it does not seem that any particular treatment would have been beneficial as the underlying process was exceptionally rapid and severe.

4.3.2 Summary of correspondence Dr Gross and the patient's mother

The patient's mother writes to MG on 6 November 2025. She explains why she believes that ornithine transcarbamylase (OTC) deficiency leading to hyperammonaemia is the only explanation for the patient's rapid decline. This is because the autopsy found no primary brain disease, ammonia is uniquely toxic to the brain, hyperammonaemia causes the type of fatal cerebral oedema the patient experienced, and negative genetic tests do not exclude OTC because of de novo mutations.

MG replies on 20 November 2025, acknowledging that OTC deficiency on the balance of probabilities could well have been an isolated example of an adult OTC deficiency with cerebral oedema. However, he emphasises that he is not a genetics specialist and cannot reach a definitive conclusion. He introduces exercise-related encephalopathy as another possibility, referring to the patient's cycling earlier that day.

In a further letter dated 19 December 2025 MG notes that the likely diagnosis sits between OTC deficiency, an undetermined encephalopathy, or a possible exercise-induced encephalopathy. He notes that the molecular analysis of the donated liver tissue did not identify a genetic cause of hyperammonaemia, though he stresses that this is outside his expertise.

In a subsequent letter of 7 January 2026 the PM describes the alternative diagnoses suggested by MG as speculative and unsupported.

On 22 January 2026, the PM responds to a case report MG had cited and provided, (*Hiding in plain sight: A Case of Ornithine Transcarbamylase Deficiency Unmasked Post Liver Transplantation*), pointing out that it reinforces her position. The case describes a previously

healthy young person who collapsed with nausea, headache and cerebral oedema—later found to have unrecognised OTC deficiency because ammonia was not measured. She asks MG and the Trust to identify any credible alternative diagnosis.

5 Other concerns not addressed in this report

Inquest process - that a neurologist was not present at the inquest. That the coroner did not take into account the mother's memorandum outlining areas to be addressed by the coroner.

6 Summary of learning

The Trust's learning from this case includes:

The importance of considering the range of possible diagnoses for headache and acute confusion (or any other clinical presentation), and in particular the importance of ruling out those conditions for which time-limited emergency treatment might be required.

The importance of following national guidance on criteria for further investigation of patients with headache (in the emergency setting, generally by CT scan).

Discussion of the patient's initial CT scan at a radiology learning meeting. The report of the external neuroradiologist has since been shared with the divisional clinical director for imaging, the author of the discrepant report (on the first CT scan), and the author of the report on the subsequent MRI scan.

The importance of initiating appropriate early investigation of acute hyponatraemia (low serum sodium), and treatment in line with best practice (a new guideline on hyponatraemia has been added to the Trust's Medical Emergency Document Library, to support clinicians treating patients with hyponatraemia).

The importance of a blood ammonia level in patients with encephalopathy without obvious alternative cause (intoxication, infection, trauma or vascular) in whom obvious causes have been excluded. The Trust has now included ammonia in the acute confusional state guideline in the Medical Emergency Document Library. This also links to related conditions including hyponatraemia.

The importance of collateral history from patients' families, and so facilitating contact between them and clinicians on the Acute Medical Unit (this was substantially a problem associated with the restrictions of the pandemic).

Involvement of senior clinicians in conversations with family when a patient has deteriorated unexpectedly.

The importance of providing full explanations as early as possible.

<u>Cultural learning</u>

It is recognised that cognitive error contributes to most diagnostic error in emergency healthcare. The Trust's serious incident report noted cognitive biases as a potential contributory factor to the problems in care identified. Morbidity and mortality reviews in Emergency Services have included consideration of the potential role of cognitive bias. Among the many strategies to mitigate the impact of cognitive bias and diagnostic error, the following is highlighted. At an individual level, there has been discussion of common cognitive biases and the circumstances in which they occur. Junior doctors in particular have been encouraged to use diagnostic pauses to ask questions like 'What could I be missing?' and 'If so, what would the consequences of that be?', to encourage critical thinking; and to take steps to manage their cognitive load to facilitate this. It is recognised that individual awareness of cognitive bias alone is not sufficient to manage the impact; and the Trust has

systems in place which it is hoped mitigate the impact of cognitive bias, particularly among less experienced clinicians.

Examples of these systems include guidelines for diagnosis and treatment (such as in the Trust's Medical Emergency Document Library), and templates for documentation of clinical information / checklists on the Trust's electronic healthcare record system - to support rational clinical decision making. Also, clinical response teams for certain presentations; and policies such as the requirement that patients' care be discussed with an appropriately experienced clinician before a patient is discharged from the Majors area of the Emergency Department.

More generally, better diagnosis is supported by developments such as improved access to advanced diagnostics; and ensuring the results of investigations are reliably received and acted on.

Catherine Mooney

Director for Quality and Safety

February 2026