University College London Hospitals
NHS Foundation Trust

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Dear Lady Young

My name is Dr Ben Killingley, I am the clinical lead for acute medicine at UCLH. Whilst I was away on secondment last year Christine Gregson took over my role, and now she is on leave. Before I continue, I wanted to say how sorry I am to hear about what happened to Gaia, I can only begin to imagine the pain and grief this will have caused you and your family.

I am getting in touch now with responses to questions you sent after the Coroner's office made contact about the potential to conduct some further tests on samples obtained from Gaia. My responses are as follows.

- **Why now? The results might have been helpful for the inquest?**

After the inquest, the coroner received an email from a doctor (an endocrine specialist) who had seen the press coverage of the case. They suggested that a metabolic disease called porphyria might have been implicated in what happened. This was passed to Professor Sheaff (the pathologist) who commented "*I think this ought to go to the clinicians really in case they have residual samples to test for the enzymes. I have looked up autopsy reports of this and they seem to suggest more chronic symptoms and peripheral nerve abnormalities. I don't think there is anything in the tissue I have to confirm or otherwise, except that if he's right it might be in the family and the tissue might be suitable for genetic testing*."

Following the email from the coroner, contact was made with a metabolic diseases specialist at UCLH. After reviewing the case they agreed with Professor Sheaff that porphyria was unlikely, but did suggest that a rare metabolic condition called ornithine transcarbamylase (OTC) deficiency could have been a cause of the symptoms seen in Gaia. OTC deficiency is a genetic disease that causes too much ammonia to accumulate in the blood (hyperammonemia). Ammonia is toxic when levels are too high and especially affects the nervous system.

Given the above, we wanted to see if we could do anything to confirm or refute this possible diagnosis.

- **Which tissues are you interested in?**

Blood taken at post mortem - a small amount was sent to a toxicology lab at Imperial College and it is currently stored in a freezer there. The pathologist has retained some small tissue samples fixed in paraffin that were used for histopathology examination. These are located at Bart's Hospital.

There are no samples stored at UCLH, blood samples are not kept for more than a few days.

- **What tests are you proposing?**

Testing for OTC and other related metabolic diseases can be done in a few ways, but given the limited samples we have, we think the best way is to undertake genetic testing. The best sample to use for this would be blood.

- **Who is going to undertake the tests?**

The testing would be done at a specialist centre at Great Ormond Street Hospital. Other tests could involve looking for certain proteins (amino acids) in the blood but we think the genetic testing gives the best chance of looking for evidence of OTC deficiency.

- **How much of the sample will you need? Would any remain for an independent lab?**

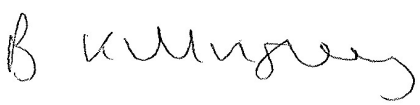
This is difficult to say until the end of the testing, but it would be unlikely any will remain given the small sample sizes that have been identified.

- **Will you share the results?**

Yes, we will share the results with yourself, the coroner and the team involved at UCLH. Importantly, if a diagnosis of OTC deficiency or other genetic disease was made, this could have implications for relatives of Gaia. It could also have implications for the recipients of Gaia's organs.

Please do let me know if other questions arise. If you are content for me to go ahead and arrange the testing described above, then please reply stating your consent.

Yours sincerely



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