‘Tiny test, huge impact’ – an essay drawing on experience of a patient encounter which illustrates the impact a pathology result can have on a patient and their family.

By Chuer Zhang

A six-year-old boy was taken by his family to the emergency department (ED) for assessment of an apparently innocuous ecchymosis on the left arm. An adventurous child, the family assumed it is likely of traumatic aetiology. Little did they suspect that this was actually a symptom of something much more sinister, and his attendance would set into motion a chain of events than would lead to the life-changing diagnosis of acute lymphoblastic leukaemia (ALL)[1].

As the news was relayed to the family, it was clear they were unaware of the enormity of the diagnosis or indeed its ensuing implications. Multiple investigations would be warranted, including bone marrow aspirates and regular lumbar punctures - daunting procedures for adults, never mind a young child. The samples were closely scrutinised in the pathology laboratory to detect any potential CNS spread[2]. Gruelling treatment would need to be administered. The physical and emotional burden would be heavy for this young child.

Incidentally, the sister of the patient was a medical student, and she immediately set out to research as much as possible about the disease. She learnt that children with ALL usually do well; males have worse outcomes; of the two subtypes (T and B-cell ALL), those with B-cell subtype have a better prognosis[1]. It was therefore a further setback when pathology confirmed the patient’s diagnosis was of the T-cell type. With this came more intensive therapy and heightened apprehension for the family.

The patient and family appeared to quickly settle into this strange new routine of daily life and friendships were made with families of other childhood cancer patients. Remarkably the boy himself appeared to take the life-changing diagnosis and its treatment all in his stride.

At the end of the first treatment block, the family were aware of importance of the minimal residual disease (MRD) results. They appreciated its significance as an indicator of response to treatment and subsequent prognosis[3], and their anticipation, and indeed apprehension, of the results was palpable.

“The MRD is high.” Each of those four sombre words from the oncologist struck their hearts like tiny little daggers. How remarkable that the course of this child’s life over the next few months hinged on the result from a minute specimen on a thin slide. The chemotherapy regimen was changed from B to C on the UKALL protocol, the most intense chemotherapy recommended.[4]

Eight months later, more encouraging results finally arrived. The patient's MRD was low enough for him to be declared "clear", and to commence the maintenance phase. The family were aware that although the course would be more prolonged, lasting two years, it was much milder chemotherapy. Their eyes brightened as they began to appreciate the full implications of this; the ability to return to school and to see his friends again, and to go on holiday as a family. From a world turned upside down for nearly a year, they tentatively anticipated the ability to savour the joys of everyday life.

Their happiness didn't last long. Less than two months later, just after his seventh birthday, the patient complained of a headache. Perhaps it was a viral infection as he had begun to integrate with other children again, they thought hopefully. A repeat lumbar puncture was performed and relapse was confirmed, with the news that it entailed more resistant disease and associated CNS spread.

A stem cell transplant and further chemotherapy was offered. His sister was the prime candidate for a match for stem cell donation. An agonising wait for the HLA results ensued and she acknowledged to staff how profoundly she wished she could be offered the chance to be part of his cure.

They didn't match.

Whilst at first devastating, via the organisation Anthony Nolan[5], a match was found in China. Stem cells from a complete stranger were flown across the world to Bristol for this little boy. Although potent chemotherapy successfully cleared the CNS disease, another wave of disappointment soon hit. He had experienced a further relapse just prior to the date for transplant. Despite initial uncertainty as to the prudence of proceeding, with a further bone marrow aspirate came the encouragement that the MRD was sufficiently low to allow transplant.

Less than two months later, still in his hospital room, the headaches returned. The pathologist examining the sample of CSF from repeat lumbar puncture was able to see cells, but were they cognisant of the heartbreak, disappointment and sorrow before their eyes as they scanned the slide and confirmed the presence of leukaemic cells indicative of yet another relapse?

This time, there was no curative treatment on offer. Palliative radiotherapy and chemotherapy were administered. The long journey that had begun fifteen months before was now reaching its conclusion. Its burden seemed to hang particularly profoundly on the sister of this little boy, who was so intimately involved in his case, from researching the disease at time of diagnosis, to being present when promising results were discussed, to the devastation of that final lumbar puncture.

That sister is me.

Biopsies aren't simply pieces of tissue under a microscope. Although mere milligrams, they weigh much more heavily in the hearts of patients and their families. In a small way they encapsulate hopes and dreams, vitality and sickness, life and death. Life has many dichotomies and these small glass slides manage to embody an element of this. The role of the pathologist was intimately woven into the sombre tapestry of my brother's illness.

As medical professionals, it can be challenging to understand the impact such a tiny test under the microscope can have on the treatment and ultimately the lives of a patient and their family, unless one has experienced it first-hand. Our hopes were pinned upon each result, every one of them sending our spirits rocketing or tumbling, and ultimately altering our lives forever. Whilst specimens can be artificially preserved, how I wish my own brother's life could have equally been sustained. My brother's journey may be over, but our memories will always remain.

References

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