

**What are the specialist local conditions, particularly genetic, that occur in the UK and require specialist tertiary care, and are there any differences between the French population (which I'll have seen in my other elective with the Necker Institut in Paris) and our English population.**

At the turn of the millennium, I read human genetics Dublin's Trinity College. The 4-year course taught me a great many things, and a few of them I still remember with a mixture of curiosity and wonderment: molecular watchmakers<sup>1</sup>, iridescent parasitic surface proteins<sup>2</sup>, simple organisms giving their more complex descendants an elaborate run-around<sup>3</sup>, neural development co-ordinated by hair stimulants<sup>4</sup>. I also remember a fair bit of our teaching on retinitis pigmentosa - a hereditary, degenerative disease of the retina which leads to significant visual impairment. The inheritance pattern of 'RP' is varied, including autosomal dominant and recessive, X-linked and sporadic. RP was then known to be particularly Irish in incidence and my lecturers were obviously moved by a sense of compassion for the local populace, much akin to the French efforts on *Listeria*<sup>5</sup>. I've searched in vain for data sets comparing the incidence ratios for diseases across countries. There are some disease-specific studies as well as some aggregated data on cancer incidence but very little is available which compares incidence across multiple diseases. I researched the subject with my colleagues at GOSH: their views were that GOSH serves such a heterogeneous and geographically dispersed community that the resultant patient demographic is unlikely to be at all quirky.

**In light of my experience at the Necker Institut in Paris, I'll be better placed to see how referrals and shared care for complex chronic diseases are managed between two leading health systems.**

This is quite complicated, GOSH serves as a centre of excellence with referrals from well beyond the national boundary, and includes many private patients. However, my experience of the NHS allowed me to follow the management of these patients into the community and understand the narrative of their clinical course far better than at the Necker. It seemed that community nursing and general practice was held in high regard by many practitioners as an essential part of the multi-disciplinary team. It was not so much that the Necker disparaged their primary care colleagues so much that they didn't mention them. On a few occasions clinicians noted (both to each other and to patients) that their primary care health professionals will likely not have ever seen or heard about this type of infantile epilepsy or that type of bone marrow dysplasia. It was explained that, while these conditions may seem common on the ward and well known to all the staff involved, it was unreasonable to expect such familiarity across the health service. In sometimes seemed that the parent had become a lay specialist in the field, like a member of the jury, and would eventually become far more familiar with the condition than the primary care clinicians caring for their kid. I met a very nice nurse specialist employed by a charity for a unique condition with only ~ 250 patients in the country. She knew the condition very well and provided a great

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<sup>1</sup> In the absence of a complete fossil record, molecular phylogenetics requires some understanding of the factors which determine the rate of DNA sequence evolution. Searching for these factors and quantifying them is hard work indeed. The researcher styled himself a 'watchmaker' – if he could calibrate the factors, he'd have made the watch.

<sup>2</sup> Researchers hoping to treat and cure African Sleeping Sickness were both infuriated and amazed by the elegance with which the *Trypanosome* parasite managed to change surface proteins.

<sup>3</sup> It was with bafflement that we were told about the ability of *Toxoplasma gondii* to make rodents impervious to the smell of cats. How could an organism without a nervous system manage to so subtly manipulate that of one far more advanced.

<sup>4</sup> Follicle-stimulating hormone – which sounds like a treatment for male-pattern baldness - seemed play an inordinately large role in the coordination of that most complicated of things: the growth and development of our nervous system.

<sup>5</sup> Apparently, France is ground-zero for *Listeria* infections.

deal of support for the families, but it struck me that there would be no role for her without the charity. As the fragmentation of the health service into clinical commissioning groups further focuses resources at the more common conditions, rare conditions are deprived of the benefit of collective medicine. I recently read some of the neurosurgeon Henry Marsh's *Do No Harm*: he lamented the discrepancy between the size of the US and the UK – as the former is around 5 times larger than the latter, in some ways specialists can be 5 times more focused on particular conditions, 5 times more familiar, 5 times more proficient.

**What are the most significant differences in provision of haematology, oncology and neurology services to the population which are served by this hospital in contrast to the UK as a whole?**

Are there any particular treatments available to patients around GOSH that would not be available to other parts of the UK? The answer to this seemed to be a fairly straightforward 'no': if a patient needed GOSH-level treatment and they lived with the UK, they would be brought to GOSH – sometimes this provoked hardship on the patient and family due to the distances required to travel. I remember that the parents of a Scottish patient were quite different in their attitude to staying or leaving. The child had a post-operative pseudomeningocele and the results of some cultures were still awaited. Although the child was well, there was a risk of catastrophic infection. The stress of the situation was evident and their responses completely understandable: one wanting desperately the safety of the home, the other the safety of the tertiary hospital.

**I'd like to learn how clinicians, in particular those with small children at home, maintain emotional stability without becoming distant from their patients. As I'm hoping for a career in paediatrics, I'm quite wary of either becoming very upset, which would not help me or my patients, or walling myself off from them, which would diminish the doctor-patient relationship.**

When a child deteriorates, how does one maintain the emotional distance required for sufficiently objective and rational thinking? I found it quite difficult at times. There was one child that I saw on return to the ward from having had his bone-marrow aspiration. He was coughing quite dramatically and his saturations seemed to be dropping. There was concern that this was either a reaction to the anaesthetic procedures (eg laryngospasm or reactive stridor) and it was key to learn if he was suffering from a current upper respiratory tract infection. If so, and he was only coughing as a consequence of the recent illness there was much less reason to panic. As his mother didn't speak English, we struggle to ask her if he had a cough before his operation. It seemed she didn't understand, and thought we were asking why he was coughing now. She looked perplexed and quite worried, possibly wondering if we knew what we were doing. The fear in her eyes affected me, and I struggle to think how we could more effectively ask the question.

There was another patient who affected me quite powerfully - a young boy with Fragile X syndrome and autism. He was a tremendous handful for us, constantly moving, very self-directed, impulsive, reckless, needy and noisy. We only needed to interact with him for about 2 hours – I could only imagine the difficulty the parents faced, particularly with a healthy young toddler at home

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