My introduction to Lowe Syndrome by Dr Anthony Norden

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I met my first patient with Lowe syndrome in 1985. We were studying the proteins found in the urine of patients with a rare group of kidney diseases called 'Fanconi Syndromes' and Lowe Syndrome is one type of these. At the time I was a junior doctor in the 'Senior Registrar' grade. The work was in collaboration with Professor Oliver Wrong and Dr. David Brenton at University College Hospital (UCH). We were interested in comparing the proteins in the urine of patients with different forms of Fanconi syndrome because we felt this could provide a clue to why the kidneys of these patients so frequently fail.

Because proteins in urine are unstable it is important to obtain fresh specimens and cool them quickly to prevent the proteins being destroyed or altered. So one Saturday afternoon, with an ice box in the boot of the car, I set out to the South London suburbs to meet the family with a son who suffered from this disorder. It was an unforgettable experience.

The care, given in his own home in modest circumstances, which was lavished on this severely handicapped boy suffering from several physical and psychological problems was very moving. Although I had met many severely handicapped children during my training, these had always been in a hospital setting and to see the intensive care provided in an ordinary home was staggering. (It is worth noting that by no means all patients with Lowe syndrome suffer from such severe disabilities as I came across that afternoon).

The parents had kindly arranged collection of the urine and after a brief 'thank you' I returned to the laboratory at UCH. There the urine specimen was divided up and and frozen in an ultra-cold freezer to preserve the proteins for later study.

Interestingly we discovered in the urine of that patient (and other with Lowe syndrome as well) a previously unrecognised protein. And as luck would have it, and as it often the way, this led about 15 years later to another group identifying yet another protein which normally controls protein movements within the kidney.

This sort of work has led to a much better understanding of what proteins occur in the urine of patients and even some idea of 'why' this happens. We are, of course, only one of a number of groups throughout the world interested in these disorders. Unfortunately we still don't really understand why the kidneys start to fail in these disorders (although we have ideas) – much less can we intervene to stop or slow down this process. Urine is now studied with techniques such as 'mass spectrometry'

in which proteins are shot into a machine a little like an 'atom-smasher'. Such technology did not exist in the protein-world of 1985.

Where is this all going and how will patients stand to benefit? Groups world-wide are well-set to put together a detailed molecular picture of the events in the kidney; at the moment these are still obscure. Perhaps closer to the needs of patients, how will this help clinical care? It is simply too early to know. Might there conceivably be ways to slow or even stop the damage to kidneys in Lowe syndrome? It is simply too soon to say. The pace of research judged by production of research reports (papers) is fast; applying this knowledge to clinical care is slow but will surely come.