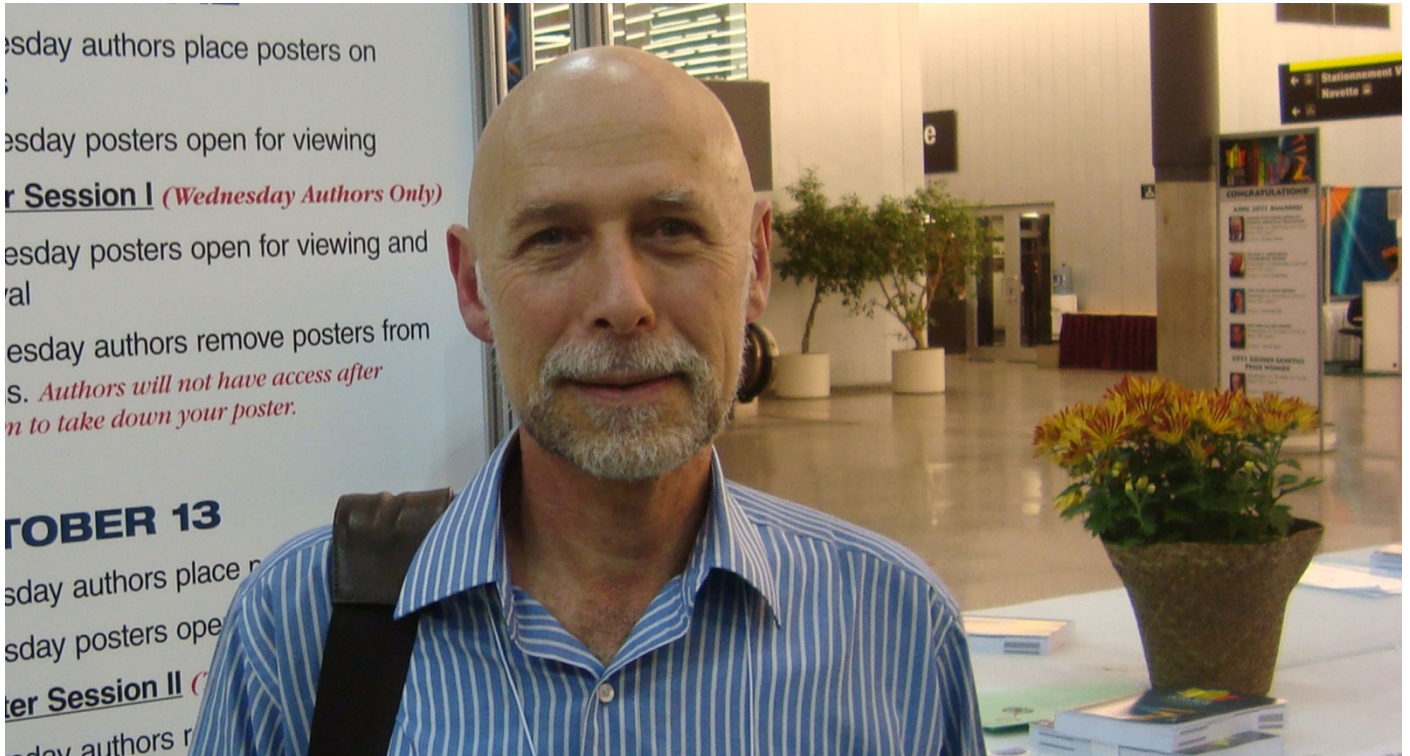


# Eric Haan



## Personal Details

Name	Eric Haan
Dates	Born 1948
Place of Birth	Melbourne, Australia
Main work places	Murdoch Research Institute, Melbourne; Adelaide Royal Children's Hospital
Principal field of work	Clinical Genetics

## Short biography

After training in Paediatrics in Melbourne, he joined David Danks for Medical Genetics research at the Murdoch Research Institute, before moving to Adelaide as Head of Clinical Genetics.

## Interview

Recorded interview made	Yes
Interviewer	Peter Harper
Date of Interview	14/10/2011
Edited transcript available	See Below

## Personal Scientific Records

Significant Record sets exists	-
Records catalogued	-
Permanent place of archive	-

## INTERVIEW WITH ERIC HAAN, 14/10/2011

**I = Interviewer (Peter Harper)**

**H = Eric Haan**

**I It's 14th October 2011 and I'm talking with Professor Eric Haan from Adelaide at the International Human Genetics Congress in Montreal. Can I start, Eric, by going right back to the beginning and ask where and when were you born?**

H Right, I was born in Melbourne in 1948 and grew up in Melbourne.

**I Which particular part of Melbourne?**

H Well, the suburb was called South Yarra, which is close to the inner city, and it was/is a reasonably well to do suburb, but it abutted on quite a poor suburb, and there was a short laneway connecting the two and I used to enjoy mixing with people and children of various styles and different homes we could run in and out of.

**I Did you come from a family which had a medical or scientific background at all?**

H Not at all. My mother, I think, in her early years did various secretarial jobs, but didn't work when the family came to Australia. My parents came from Hungary, from Budapest, my mother having a Rumanian/Hungarian origin, my father purely Hungarian. My father went into his father's printing business, which printed office stationery and so on for businesses around Europe. And so there was not a sniff of a medic lurking in the family tree that I was aware of.

**I Were there any special factors that you can think of that took you in the direction of medicine?**

H Not really. I don't really know what made that happen, Peter. I think that at the end of my high school years, I had what was called a reserved place in the science faculty at Melbourne University. I did a second year of matriculation, having done all sciences in my first matriculation year, and did history, economics and languages. And during that year I decided that perhaps I'd rather do medicine than science, but not with any clear vision of why, in any way; I'd always enjoyed almost everything I'd done, and if you look forward in time to the various specialties in medicine, I continued to enjoy almost all areas that I was exposed to in my training, but in that second year of matriculation I decided to do medicine and changed over. And in doing medicine chose to go to the young, upstart university at Monash, where many of the grounds were still slightly muddy because the builders had just left, and found it to be a very interesting university because it had attracted, as with many new universities and hospitals, as we've seen in Adelaide, young, energetic people to head the various departments, and it had a very fresh esprit de corps which doesn't exist perhaps to the same extent in older universities. And so I chose Monash University because it seemed to have a very good course and I was, just by personality, inclined not to go to the, in Melbourne's case, established university, ...[laughs]

**I Was the medical school at the Clayton Campus?**

H Clayton Campus; exactly. That's where it was, so I had to go out there every day in a battered old car driven by a friend.

**I And what year was it that you qualified?**

H I qualified in medicine in 1972 and did my intern years at the Alfred Hospital, again in Melbourne. And so that was 1973/4, and in 1975 there was a great opportunity to undertake

further training in neurology at the Alfred Hospital and I was encouraged to do that. But again, for some strange reason, I decided that paediatrics was more attractive to me, and went to the Royal Children's Hospital to train. And it was there, in my second year of paediatric training, that I did a rotation that combined neurology and genetics. And just as an aside because it's of historical interest: the head of neurology was Ian Hopkins, who was involved in the process of describing Pitt-Hopkins syndrome; David Pitt was also at the hospital at the time and so Pitt-Rogers-Danks syndrome as well as Pitt-Hopkins syndrome. Several syndromes were described at that time. David Danks was the head of the genetics unit and I found him a very inspiring individual and it was his influence, as so often happens, that led me into genetics. I think we had a personal affinity; we had a similar, I guess, academic approach to, well a curiosity and a keenness to really make, if you like, research use of every patient we saw; anything unusual was interesting. David was interested in academic things, thinking about patients, developing ideas about them, hypotheses, and pursuing them; and I found that to be exactly what I was interested in as well. And when my second year of training was over I applied to join David and train in genetics.

**I At that time was the medical genetics department, was it very orientated towards metabolic diseases?**

H Yes. As you know, David Danks was a neonatologist to start with, and at the time the metabolic and cytogenetics laboratories were part of the main pathology department. David had taken an interest in the clinical management and diagnosis of metabolic disease and was starting to have a significant influence on the running of the metabolic laboratory. So metabolic disorders were, I guess, an important component of the department. But it was broader than that. I think David, having trained with Victor McKusick, was interested in connective tissue disorders, and dysmorphic syndromes and multiple malformation syndromes, and so the exposure was broad at that time. But there was a big focus on inborn errors. David's own research activities with Dick Cotton focused on PKU and trying to understand the genetics of it and how best to manage it. David ran the PKU clinic as well as the metabolic clinic and did that together with David Pitt. So yes, that was certainly a focus on metabolic disorders but there was a lot more happening as well. David Sillence had been training overseas and came back with an interest in osteogenesis imperfecta and bone dysplasias in general. And then Les Sheffield came back and developed his interest in chondrodysplasia punctata and the mathematical analysis of pedigrees, Bayesian analysis and so on.

And John Rogers was there at the time. His interest was in general dysmorphology, bone dysplasias and genetic counselling. He had quite an interest, really, in in-depth counselling, which he developed subsequently to the extent where he became a counsellor working independently, bordering on psychiatry, in terms of what he was doing. So that was the environment. The group was still very small at the time. And around the time I got there, I don't know exactly which year it was, David Danks took on the Stevenson chair of paediatrics in the University of Melbourne at the Royal Children's Hospital, quite overtly to make use of the influence that came with it to develop genetics. David in fact delegated the teaching of students, other than a certain amount, to a colleague called Max Robinson, and Max did most of the teaching and David really, in the professorial chair, used his influence within the hospital to enhance genetics as a medical speciality and also to develop the birth defects research group.

**I When you say he delegated the teaching, do you mean the general paediatric teaching?**

H Yes, the general paediatric teaching. Yes, that's right; exactly.

**I So at least, was there another paediatric chair or department or -**

H No, that was the professorial department and I think David was being quite strategic. I think

once you became professor you sat on all the important committees and the board of the hospital, and one could develop relationships and convince people of the importance of genetics and the need to develop it. And I think in that he was very successful.

**I At that time was, did you have medical genetics clinics or were they still all mixed in with paediatrics?**

H No, by that time they were just purely medical genetics clinics. In my time we didn't have any combined clinics. I don't know, at what point the change came. It's an interesting piece of history that I'm not aware of, when the transition occurred, but certainly by the time I was there in 1975, there were no mixed paediatric genetic clinics.

**I What about outlying clinics? Had they started?**

H Yes, but only to the metropolitan hospitals. I think it may well have been after, as I think about it, I don't think we did any outreach clinics at that time in 1975/6. In fact when I think about it, that would have come significantly later as the unit developed further.

**I And what about adult disorders? Were they covered at all, or not really?**

H Only accidentally. [laughs] As you saw the children, obviously you had to consider, for dominant and X-linked disorders, that there were parents for those children, and extended families, but there was a heavy emphasis on paediatric genetics and most of the activity was in the neonatal nursery and other wards of the Children's Hospital and the hospital outpatient department. Obviously the newborn screening programme was going and samples came in from a range of hospitals, but yes, there were no outreach clinics at that stage; that came significantly later.

**I What about your own interests at this point, when you were in Melbourne? Did you have a particular research area you took up?**

H No, not really. The area I moved into fairly quickly was inborn errors of metabolism. I definitely followed David into that and under his tutelage, gradually learnt more about them. And I guess any research I did was clinically related research; it was related to what we found in urine metabolic screens and in unusual patients. That reminds me of some of the things we did in the past, and one wonders, in retrospect, whether they were quite appropriate. I remember siblings with nonketotic hyperglycinemia; we'd found that strychnine blocks glycine receptors, so we gave the children strychnine to block the effect of the excess glycine in the brain. It didn't harm them at all; in fact it didn't do anything. But that exemplified David's interest in pursuing an idea or a hypothesis even in the clinical setting. And I can remember him, for a whole range of undiagnosed inborn errors, thinking "Well, we haven't got a diagnosis and we've come to the end of the standard therapeutic armamentarium; are there any clinical features in the patient which would suggest that a certain class of drugs could be tried as an experiment of one?" And you know, he might have a patient with dystonia and, "Right, let's manipulate the dopaminergic system and find drugs that switch it on, drugs that switch it off" and did short-term clinical trials to see what would happen.

David certainly had individual cases, difficult cases, where he had thought about what might be going on and not being sure, but getting to a point where he considered it appropriate to try to manipulate the system, for better or for worse, to see whether there were improvements or deteriorations. Whatever the outcome, it gave you the opportunity to think about what might be going on. Fortunately we didn't have too many deteriorations, if any, but that gives you the flavour.

David was particularly interested in the concept that there might be inborn errors of neurotransmission or neurochemistry. And he interested me in that. And based on that interest, supported me to go overseas for three years. The concept was that the principles that applied in systemic inborn errors of metabolism might also apply in the brain. That was a

time when we didn't know so much about neurochemistry or brain metabolism and so it was unclear what one could do to try and sort them out; the idea was that there might be metabolic disorders that were limited to the brain and access would be difficult for diagnosis and treatment.

But we certainly were enthused about it and I applied for a fellowship to go overseas. I was successful in getting that, and went to the Institute of Neurochemistry in London and worked there for a couple of years on Alzheimer's Disease, with David Bowen, in Alan Davison's department at the Institute of Neurochemistry. We also worked on ageing. It was really an opportunity for me to try to develop some level of understanding of neurochemistry, because I had no neurochemistry or neuroscience background, and to give me some idea of the technologies that applied and perhaps to develop ideas that would lead on into the clinical arena.

**I What years were those that you were in the UK?**

H It was 79/80 and at the end of that time I had an opportunity to go to the Salk Institute to work with Maxwell Cowan. Max headed the neuroanatomy laboratory at the Salk. I was there for a year and we made monoclonal antibodies to neurone-specific antigens, particularly to neurone-specific enolase and also tried to culture hippocampal cells. [laughs] And you can imagine this medical practitioner with no qualifications at all in science. Anyway, we managed to make a monoclonal antibody, probably by luck rather than anything else, and were able to publish that. Hippocampal cultures, [laughs] which I think it was very ambitious to suggest that I do, weren't so successful. But it was a wonderful year, obviously the Salk Institute had several Nobel laureates including Francis Crick, and it was a wonderful environment.

**I While you were in Britain did you make any links with the genetics community; medical genetics community at that time? Or weren't you really aware of them?**

H Not really aware of them. I didn't make any real links with the clinical genetics community.

**I What happened next? Did you go back to Melbourne?**

H Well, I went back to Melbourne to a position that was a mixture of clinical work, which was again in inborn errors of metabolism, and research activity that was related to neurochemistry. It was at an early stage and looking back, I was extremely green and it's not surprising that in the end I didn't make a go of it. I made some monoclonal antibodies to the brain hydroxylases, phenylalanine, tyrosine and tryptophan hydroxylases, and looked at where those antibodies went in the brain. And again, it was related to David's interest in phenylketonuria and phenylalanine hydroxylase. We did some work on CSF samples from children with unusual neurological disorders, but our capacity to analyse, or even know what to analyse, was severely limited. And we also set up an assay for glutamic acid decarboxylase, thinking as a producer of GABA it might be an interesting enzyme to study in a range of circumstances. But I wasn't very good at setting up biochemical assays and although I'd had a certain amount of laboratory experience by then, there was nobody really to train me; it was self-training and enzymology did not come easily. However, the techniques I had acquired overseas that I did feel reasonably competent in, like the antibody studies and immunohistochemistry, seemed to go reasonably well, but the straight biochemical assays were a bit tricky for me.

In the clinical arena we tried to identify patients who might have inborn errors of brain chemistry, uniquely brain chemistry, and certainly identified a lot of patients but made no progress whatsoever in uncovering the basis of their disorders. At that time you could obtain CSF but could only look at amino acids, organic acids and perhaps pterins. There were people in Sydney who could look at neurotransmitter metabolites for you, but there was nothing that we really sorted out. There was one group of patients with lactic acidosis and manifestations

largely limited to the brain and it was possible to define some phenotypes in several patients with those abnormalities. It's worth mentioning that around that time, when I'd got back from the UK and the US, a wonderful biochemist/clinician, Gary Brown, and his wife, Ruth, who was a tissue culturist, had joined David's group. Gary was very interested in lactic acidoses and mitochondrial disorders. This was in the days before David Thorburn had arrived. Gary and I worked together to try and sort out some of these patients, but at that time it wasn't very successful. That would have been 1982/3.

**I Was this the time also when David was very interested in copper and metabolic defects like Menkes and things of that type?**

H Yes, it was. His interest had started a little earlier, but yes he was extremely interested in copper and Menkes disease. The anecdote about him, I don't know whether you've heard it from others, is that the inspiration came to him in the shower and so [laughs] David would traditionally say if he needed to have an idea he'd better hop into the shower. He was thinking about copper deficiency in Australian sheep and made the connection with kinky hair in Menkes disease; the penny dropped as he was soaping himself in the shower [laughs] - his Eureka moment! Yes, he had a real interest in copper. He worked with Jim Camakaris at the University of Melbourne who did the copper assays. Jim had been in David's group at the Royal Children's then moved to the University of Melbourne, and did a lot of laboratory work in relation to the copper studies.

**I Now, how had your kind of career progression been going on during this time? Had you got to a consultant post by now or were you at a lecturer/research fellow, that sort of stage?**

H I think you would have called me a junior consultant by then but I had not completed training in clinical genetics. The genetics training system in Australia, until 1994, was that the Human Genetics Society of Australasia did the accreditation, and after 1994 accreditation was through the College of Physicians, like other specialities. It was only towards the end of my time in Melbourne that I had done what was necessary, and so I was a junior consultant in paediatrics rather than clinical genetics at that stage, having finished the paediatric qualifications and being well on the way in clinical genetics and nearly finished.

**I Can I just check: which year was it that medical genetics became a full clinical specialty in Australia?**

H That is a very interesting question because [pause]... I honestly don't know the answer to that because it was a bit before my time. I was recognised by the Human Genetic Society as a clinical geneticist in 1987 but the process of recognition had been implemented before then.

**I But it was already a full specialty.**

H Well, it was a recognised specialty after 1993-4, recognised by a national body called NASQAC, which was a group that determined whether you were a consultant medical specialist for Medicare billing purposes, rather than that you were qualified in clinical genetics. And they had accepted clinical genetics training as equivalent to the other consultant specialties in terms of billing. It was clearly a full medical speciality from 1994, when training fell under the aegis of the Royal Australian College of Physicians.

**I What year was it that you moved to Adelaide?**

H 1985

**I And what was the background to that after being all your life in Melbourne really?**

H I think there were several elements to it. To be frank, my research activities in Melbourne had not gone as well as they should have and I think David had great expectations for the work. But also, I had come to a view that I was not destined to be a research person; that I enjoyed clinical work too much. And I think David was keen to have clinician researchers with a strong

laboratory base, although I'm sure he would have been flexible about the sort of research. My impression was that he saw me as someone who he hoped would take on a largely research career and I guess my assessment of it was that I would rather be a clinician fundamentally. And when the job in Adelaide came up, I applied for it.

The clinical genetics unit in Adelaide had been started in 1979 by Les Sheffield, and he, in 1984/5, had reasons for wanting to go back to Melbourne, having come from Melbourne. That created an opportunity in Adelaide, I applied for the job and was successful. I suspect I was the only applicant for the job because there weren't many people with the necessary qualifications to take it on, but it was, in retrospect, a wonderful decision from my point of view. My personality is that I'm relatively, I like to think, [laughs] shy and private, and to go to Adelaide as the only clinical geneticist gave me opportunities and experiences that I might not have had if I had stayed in Melbourne. At that time the Adelaide unit had one clinical geneticist and one co-opted genetic counsellor. Again, as an historical aside, that counsellor had been selected by what was still then 'the matron' of the hospital rather than the 'head of nursing'; she'd gone down to the casualty department and tapped a particular nurse on the shoulder and said, "I think you'd be good for genetics. You start on Monday". [laughs] Anyway... and we had a secretary. So it was a very small department and for someone with my personality, I was forced to interact with many other people, to be much more of an advocate than I ever would have been in a different environment, and I think the experience changed me in terms of the way I behaved and that sort of...

**I Yes. In Adelaide was Grant Sutherland already established in cytogenetics?**

H Yes. Well, it's true that Adelaide was a most wonderful place to go at that time, and the established research groups were one of the great attractions. It wasn't just that I was leaving the Melbourne scene, it was that I was going to a centre where one knew that there were a number of excellent research groups. And one of them was Grant's group. Grant had started as a hospital scientist running the cytogenetics unit and over time did more and more research, recruited John Mulley to start working on X linked mental retardation and gradually increased the size of research activity so that ultimately it, not quite dwarfed, but certainly was much larger than, the clinical service activity in cytogenetics. But the two went beautifully hand in hand under Grant's guidance. Grant was very flexible and it was a time when there were none of the tight boundaries that now exist between research and service in terms of finance, and department heads had much more authority to make administrative and financial decisions; sometimes the research would be subsidising clinical activities and sometimes clinical activity would be subsidising research. The two were totally integrated.

Grant and I had a very good personal relationship, so the relationship between the 'clinical unit of one', and Grant's research and laboratory service activities, created a wonderful opportunity for me. I was dragged along on Grant's coat tails in many of the areas where I subsequently was involved in publishing and was able to provide him and his research team with interesting cases and families.

And he was very generous about that; he had the philosophy that anybody, whether clinicians or otherwise, who contributed materially to papers should be authors and... I don't know whether his attitudes would meet the current guidelines for authorship but that was his view. So it was a wonderful place to work, because you were automatically sucked into a lot of research activities, particularly as the only clinician. And you saw all the patients; anybody who had anything unusual or exciting/interesting was your patient, you see, and so it was a unique situation.

There was also Tony Pollard and John Hopwood, and John obviously has gone on from strength to strength in research, working on lysosomal storage disorders. And there was a good group doing inborn errors of metabolism pretty much as a service activity, with Evelyn

Robinson. She and Tony were interested in expanding antenatal screening, as well as doing neonatal screening, and so I was there at a time when Adelaide, which had a long history of doing antenatal screening for neural tube defects using alpha-fetoprotein screening in the second trimester, started second trimester screening for Down's Syndrome.

And so that was what was going on in the years soon after I went to Adelaide. It was also a place where all the complex questions about teratogen exposure in pregnancy were directed to the clinical geneticist, and I had to learn about that. So one had a very broad exposure to clinical genetics and to two extremely strong academic groups: Grant Sutherland's and John Hopwood's. It was an interesting and exciting place to work, albeit being a relatively small centre.

**I Over the years you've been in Adelaide, is it 30 years now?**

H Not quite, Peter. [laughs] I think if we work it out it's 26 or something like that.

**I I mean what would you feel are the main things which you've achieved there?**

H Well, gradually we've built a much bigger department, which now has a significant number of clinical geneticists, genetic counsellors and administrative staff, having started with, as I said, one clinical geneticist, one counsellor and one secretary. We managed to create a paediatric, prenatal and general genetics unit with about 3.6 FTE clinical geneticists and 3 or so genetic counsellors and a familial cancer unit with 1.6 clinical geneticists and 3.5 genetic counsellors. So there's been the achievement of building up the service. By the time I got there Les Sheffield was already doing outreach clinics, both in the Northern Territory as well as to two country centres in South Australia; those two centres are the only large centres in South Australia, and I continued that. Outreach clinics to hospitals within Adelaide had also been started, and I continued that. I would say that I was able to build and extend the relationship with Grant Sutherland and that particular group, which I think was very useful as cytogenetics and molecular genetics developed and there was a very close working relationship with the laboratory for the clinicians. And Grant was very good about seeking clinical advice and responding to it, as he implemented technologies in the laboratory.

**I How about links with genetics across Australia, because it must have been quite isolated in a way in Adelaide in terms of other clinical geneticists, I mean, had that already, had the Australasian Genetics and Medical Genetics Society already built up? Or was it still at a very early stage?**

H Clinical genetics was still at a relatively early stage but was certainly in existence, with each of the Australian states and New Zealand a fiefdom on its own. But people had a sense that there was a need for a national body and the Human Genetics Society of Australasia was formed in 1978, a year before Les Sheffield established a clinical genetics unit in Adelaide..

**I I do have something on that that I think Grant Sutherland wrote.**

H The national body was well established by the time I started in Adelaide, and the annual meeting was the opportunity for everybody to get together and they did. It was at a time when air fares in Australia were fiendishly expensive, notoriously expensive, and so there were issues of financing, but certainly we met every year, and there were, as at this meeting, endless committee meetings of various groups trying to address things at a national level, compare notes and so on. But I think people were not unhappy being in their states and doing their own thing. Certainly there were differences in the way people practiced and structured their services, which could sometimes be a little idiosyncratic. For example, in South Australia, service decisions were based on the opinions of one person [laughs] and pretty much the same was true in the other small states. In Melbourne, David Danks had implemented a centralised clinical genetics service that provided outreach clinics and it is worth saying that most of the states took the Melbourne model. New South Wales, because of the much larger



population, and two children's hospitals and one adult hospital where there was an interest in genetics, was much more fragmented from the very beginning and remains so, while the less populated states, Western Australia and South Australia, and one of the bigger states, Queensland, remain centralised. Victoria has been somewhat decentralised in the last couple of years as the result of political decisions. But there certainly was, I think, the Danksian model, and it was the Danksian model that had been translated to all the states, though with a greater degree of fragmentation in New South Wales than anywhere else.

**I One thing I'd like your opinion on is how the very strongly paediatrically rooted genetics in Australia has affected its longer term development long after David Danks.**

H Yes, an important issue and one we've all been aware of, Peter. In Australia clinical genetics literally grew out of paediatrics, within children's hospitals. And until quite recently, almost all clinical geneticists in Australia have been paediatrically trained; there are a small number who are adult trained, but they are uncommon. And I think that Australia has suffered as a result; it has been a completely lopsided development, and the only reason we got away with it was that the adult clinicians didn't seem to take a great interest themselves, because it would have been tremendously fragmenting to have individual adult physicians in the non paediatric centres establishing genetic activities, yet they needed to be established. But the paediatric centres did, from the very earliest days, try to address certain adult related disorders, particularly the Huntington's predictive testing that came along in the mid to late 80s. It was offered in South Australia as an outreach activity of the paediatric service. In Melbourne it was different. I think for some reason David Danks was not particularly interested in it so the Department of Psychiatry at Melbourne University undertook that particular role. In New South Wales a social worker loosely related to genetic services, undertook it.

**I Was that Betty Telcher?**

H No, Betty was in Melbourne, I think. Fiona Richards was the lady who organised it and delivered the service in New South Wales. I don't know that there was a service in Queensland. In Western Australia, and again I don't know the history well but when I first became aware of the activity in Western Australia, it was being done by a group with links to the genetic service, but somewhat independent from it. In terms of trying to get into adult genetics we, South Australia at least, only achieved funding for familial cancer work in 1994, and from that point on we were, if you like, delivering an adult related service on top of the Huntington's predictive testing. But as you know, one is always seeing adults as part of paediatric practice: a child with neurofibromatosis, mother with neurofibromatosis, whatever. But certainly adult genetics was very underdone, as we've now realised. We're trying to address it, for example in relation to cardiac genetics, but it's hard to do at a time when funding is not so easy to get. I think people are now committed to developing it, but developing it in Victoria, for example, led to a split of the clinical genetic service. I think it was driven by, or partly by, the need to develop adult services and then a political process that led to that being done in a way which split the centralised model. In South Australia and the other smaller states we haven't had that particular problem: funding has been the problem. Nobody has wanted to fragment the clinical genetics service but it remains a fact that in South Australia there are outreach clinics to adult hospitals but no resident clinical geneticists and services in the adult hospitals.

We've been trying to achieve a proper adult service for a long time and the opportunity to achieve will come when the new Royal Adelaide Hospital opens in 2016; barring financial and political crises. It's agreed and the foundation stone's been laid. Hopefully it will be built and we believe there will be room in that hospital for an adult clinical genetics service as part of the cohesive state-wide service. So we've tried to deal with it but it's been a long time coming and it's been quite inadequate because we've had virtually no impact in the adult hospitals, even those where we do clinics. Some of the hospitals are adult hospitals with paediatric

sections and we've tended to be recognised and known about by the paediatricians and receive referrals from them, but not from the adult clinicians. In the one hospital which is purely adult, the Royal Adelaide Hospital, I come and go. It's been hard to make an impact, and in particular we've been extremely concerned that we aren't able to interact with the trainees in adult medicine and attract them into genetics. And so the system is perpetuated by having genetics units that are in paediatric hospitals; the paediatric trainees in those hospitals get excited about genetics and ask to be trained and we train them. But we don't see any adult trainees that we can suck into our vortex.

**I I know one of the things which struck me on my early visits was that almost universally in Australia you do have separate children's hospitals which is now unusual in Britain, and they seem to be very separate in terms of both bricks and mortar and in terms of the whole staffing. They've led their own existence quite largely, and that must have made things a lot more difficult.**

H Ah, it makes it immensely difficult even if they're reasonably closely located. I mean the closest you get to it is in Brisbane, where you have the adult hospital, the women's hospital and the children's hospital on the same campus, practically connected by corridors, but historically their administrations were different, I'm not quite sure exactly what the situation is now, but people who work in that environment say it's much easier because the access is so easy that people do interact and the adult people are aware there is a genetic service there. And so, yes, in Australia clinical genetics developed largely in a paediatric environment in free standing children's hospitals.

Peter, the other thing that might be worth mentioning is that the first clinical geneticist who joined me in Adelaide was Graeme Suthers and Graeme was very interested from the beginning in developing adult services and in particular was keen to move into that area; he became the head of the familial cancer service when that was funded.

We also had the view, having had the wonderful relationships we'd had with Grant Sutherland and his group and John Hopwood/Tony Pollard and their group, that strong relationships between laboratories and genetic clinicians are terribly important, and we had an opportunity within the Women's and Children's Hospital in Adelaide to become part of the Division of Laboratory Medicine in 2003.

We discussed it and thought it would be a good thing to do. Graeme and I both were strongly supportive of the idea and ultimately it was put to the hospital executive, who accepted it and we moved from being a specialist department within Paediatric Medicine to being part of the Laboratory Division. We became part of the group of laboratories that were developing the technologies that we were so keen to use and we thought, hopefully not too arrogantly, that we had a bit to contribute to the laboratory developments. Within South Australia in 2008, the Department of Health decided to create a single state wide pathology service from the hospital based pathology departments. SA Pathology was established and the Division of Laboratory Medicine, including the Clinical Genetics Service, became part of a pathology entity, which was an interesting development. But we saw it as an advantage for a couple of reasons: obviously it had the advantage of linking us very tightly to the laboratories but it also placed us within a state-wide service with no geographic, age or medical speciality boundaries. We were already a state wide service but based in a children's hospital and seen as a paediatric subspecialty service and now were going into a truly state-wide service that covered the state in every area, and was not based on age.

As part of that laboratory service was for adults, as well as for children and pregnant women, we saw the change to be an advantage, as a strategic thing for the future. And interestingly there is one other group that has done that: in one of the regions in New South Wales, the clinical genetics service has become part of the regional pathology service. There are also

close links between labs and clinical services in Melbourne and Perth.

**I Did that change cause you any problems in terms of your closeness to clinicians and, you know, affect the way that you practice, especially given that a lot of Australian medical genetics had had a patient management role at times?**

H It's an interesting question, Peter. I don't think it caused us any problems in the way we practice because the environment is that we're still sited in a women's and children's hospital, because we are paediatricians by training and because we have long standing relationships with the paediatricians and obstetricians in our home hospital and across the state. I don't think it has caused us any major problems in the way we practice clinical genetics. There are of course the usual administrative issues that arise when one administrative entity i.e. the pathology service sits within another administrative entity dealing with women and children, i.e. the Children, Youth and Women's Health Service. And the lines on the floor are fairly tightly drawn about whose space is whose and who pays for what, but I don't think it's affected us at all in terms of referrals and so on. On the other hand we are yet to see the advantage of being in the pathology service because we're stuck physically in that hospital. We have tried to winkle our way into the adult hospital where SA Pathology's main molecular lab for cancer and adult related testing sits, but they have not had the space. But we hope in the new adult hospital there will be a home for us.

**I What about links with the private sector, because another thing which has always struck me in Australia as compared to Britain is that particularly in the lab services, the private sector is very much more predominant. Is that something that's affecting you in genetics?**

H Not in South Australia. It has perhaps in other states. In South Australia, clinical laboratory genetic services have largely had and maintain a monopoly. We have a single cytogenetics laboratory. There were two at one stage: there was one smaller laboratory run by Dr Judy Ford, but she decided to move into the private sector and set up a private cytogenetics lab in, I think, 1994. And in 1995 the coordinating committee for genetic services in South Australia decided that really we should, at that point, have a single cytogenetics laboratory and that's what happened. And so we have to this day a single cytogenetics laboratory in the public sector and the private sector has chosen, for whatever reason, not to compete with that, and so chromosome testing requested through private pathology services in South Australia, are currently still done by the centralised public sector service. In terms of antenatal screening, the main service is in our hospital and there is a private sector lab also offering this service. But having good relationships with that group, we've managed each year to get their data and to combine it with our hospital's data and to publish the experience of antenatal screening truly on a state wide basis. And so the private sector hasn't been a problem for us. I think that as the private sector consolidates into large pathology or health companies, and particularly as we move into the molecular era, my feeling is that that will change, and that a lot more genetic testing will be done by the private companies, which they will send their samples to a central location to get the cost benefits of high throughputs and so on. But we shall see.

**I Eric, are there any other things that you'd especially like to bring up and go over? There are lots of things we could talk about but you know, you haven't got so much time. Do you think we've covered the main things that you wanted to?**

H Yes, I think we've covered the main things. Just trying to think: I probably indicated my feelings that I found David Danks to be inspirational and I really enjoyed working with him; an extremely stimulating person to work with. And yes, I think we've already covered the main things, Peter. I just thought I should repeat that because [laughs] David was very good to me and I was certainly inspired by him and have a great debt to him.

**I Thanks very much, Eric.**

