Arvid Heiberg

Interviewed by Trine Prescott

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Arvid Heiberg was born 20.12.1937 in Oslo, Norway. He obtained his MD in 1962 and his PhD in 1975, both from the University of Oslo. His doctoral thesis was entitled “Familial hyperlipidemia with xanthomatosis”. He became a specialist in Internal Medicine in 1971 and in Medical Genetics in 1975. From 1976 to 1985 he worked as a consultant in Medical Genetics in the Public Health Department in Oslo. He was the medical director at Frambu Health Center from 1985 to 1995. During this period, Frambu became a center for rare disorders. From 1995 until 2004 he was head of the Department of Medical Genetics at the Norwegian National Hospital (now Oslo University Hospital) where he continued to work as a consultant until retirement in 2007. He held an academic position in Internal Medicine at the University of Oslo. In 2002, he was appointed Norway’s first professor in Genetic Counseling at the University of Bergen. His main interests have been genetic counseling, osteogenesis imperfecta, Huntington Disease, the neurofibromatoses, lysosomal diseases and rare disorders in general. Most recently, in addition, he has focused on medical ethics and prioritization of resources.

*TP: Let’s introduce ourselves.*

AH: I’m a medical geneticist. I used to call myself a social geneticist; by that, I meant that I do clinical work with families.

*TP: We’ve known each other for 15 or 20 years, and worked together on and off until you retired in 2007. I am a clinical geneticist. I followed in your footsteps to Frambu, and we worked together for several years at what is now Oslo University Hospital. You are pretty busy for someone who has retired but will come back to that. Let’s start by talking about your training.*

AH: I finished high school 60 years ago and went to medical school in Oslo from 1956 to 1962. I was asked recently to reflect upon my life and important decisions I’ve made. My conclusion was that I chose medicine because of personal experience. I had tuberculosis in my hip in early adolescence. I was treated operatively; hospitalized for almost a year.

*TP: How old were you?*

AH: 13 to 14½ years old.

*TP: A lousy time to be in hospital…*

AH: Yes, it was a lousy time. During one phase I felt sick. Then I was put in a plaster cast that went from my ankle to the top of my chest. It was summer. Itchy -- not a pleasant time. I spent six months in a cast and another three or four months in traction. At a time when most of my contemporaries were running around, I was flat on my back.

There was no precedent for studying medicine in my family, but this period made it clear to me that I wanted to. So, I worked hard at school in order to get into medical school.

*TP: But the experience could have had the opposite effect. You might have decided to stay as far away as possible from doctors and hospitals.*

AH: No, I became interested, fascinated. A feeling that I would describe as “call of duty” -- that one should do something helpful for others – originated in that period. As medical students, we learned to work hard. I was a receptive guy who liked to sit and study. Medical school was a formative time -- I made a lot of friends; we’ve stuck together over the years.

*TP: You met your wife then too.*

AH: Yes, my wife and I were in the same class as medical students.

*TP: This was in the 1950’s and early 1960’s. How big was your class, how many men did she have to “choose between”?*

AH: Our class consisted of 14 women and 86 men. It took some time, but in the end, I was the lucky one. Several couples found each other in our class. Some of them remain very close friends.

*TP: After medical school, you did an internship consisting of six months each of surgery and internal medicine in hospital followed by six months in a family practice setting. Where did you do family practice?*

AH: We were out west in a windy part of Norway, in a very small place. It was called “Bryggja” which literally means “the pier” -- a pier was about the only thing there. People lived spread along a beautiful Norwegian fjord, so there was a lot of traveling by boat to see patients. I remember especially a Christmas Eve when a full-blown hurricane came in cutting off the electricity for 48 hours or so. It did away with Christmas dinner.

*TP: “Bryggja”, is it close to a town with a hospital?*

AH: No. There was a surgeon about 50 kilometers away who serviced everyone in the area, about 30 000 people. He performed surgeries, delivered babies etc. If complications were anticipated, pregnant women traveled to the city of Bergen one to two weeks ahead of delivery.

*TP: This was really the good old days.*

AH: Yes, it was. The patients were truly patient then, and their expectations were modest. I recently visited the local hospital. There were about 12 consultants in various fields. Progress for patients has been enormous.

*TP: Quite a difference from boyhood in Oslo.*

AH: It was a very good time. We learned a lot. Even though I had just finished medical school, I had quite extensive clinical experience already because we supported ourselves through medical school by working part time. I had worked on internal medicine and surgical wards as well as in emergency rooms. I was an experienced version of an inexperienced doctor.

*TP: Reasonably well prepared.*

AH: Yes, reasonably.

*TP: When you were done with your internship and had become a fully certified physician, you ended up in Internal Medicine. Where?*

AH: In a city called Fredrikstad, which is 100 kilometers south of Oslo, at a quite large district hospital. I wanted to do Internal Medicine

*TP: Because?*

AH: I liked Internal Medicine. It suited my personality. I was never a surgeon -- never very good with my hands. The combination of learning and seeing patients I liked. I was lucky because the head of the department was very hard working. He managed to keep all relevant information about 120 patients in his head. We learned to work hard. It wasn’t a great academic setting, but it was a good place for learning and I liked Internal Medicine.

*TP: So now we’re talking about the 1960’s?*

AH: Yes, and in 1968 I did a year of Clinical Chemistry at the hospital in Fredrikstad. By then we had two children. A job came up and I needed something in addition to Internal Medicine in order to be certified as an internist, and I liked the head of the department. This was when the transition to automated analyses was happening; the bulk was still done manually.

*TP: A full-fledged internist, what next?*

AH: We moved back to Oslo. Both my wife and I have family here. I am born and bred through generations in Oslo. I spent some of my childhood in other cities, but what I remember is growing up in Oslo. A job opened up at the Medical Department A at Rikshospitalet, the National Hospital, traditionally regarded as “The” Department of Medicine -- the place to be. The timing was not so good research-wise because of a transition in leadership. I worked as a clinical instructor for medical students and liked it well enough, but it became a bit repetitious.

*TP: Why?*

AH: I had to repeat part of the same syllabus every two weeks with a new group of students, 26 times a year.

*TP: Was it a combination of didactic and bedside teaching?*

AH: Yes, I liked the didactic part and I liked getting the patients to talk about their experiences.

*TP: But a bit repetitive.*

AH: Yes, and then I got a call from two colleagues I had gone to medical school with, Helge Boman and Harald Torsvik. They had started to work for Professor Kåre Berg who was back from Rockefeller University in New York. He had trained with Alexander Bearn and had an opportunity to hire people. We got together. My salary continued to come from my teaching job. My clinical training was useful because Kåre Berg suggested doing something on hypercholesterolemia. My former boss in Fredrikstad had been checking everyone routinely for xanthomas and had a large collection of patients.

*TP: He had patients with lipid disorders and Kåre Berg was interested in lipid metabolism…*

AH: Yes, Kåre was interested because he had isolated lipoprotein A (Lp(a)), which was his claim to fame. He wanted to look at a possible association with premature coronary heart disease. For some reason, we dropped this project at an early stage. We went on to do a clinical study, showing that hypercholesterolemia was an autosomal dominant disorder. Markers and linkage.

I had 160 probands and collected blood from them, all their siblings, children, parents, aunts and uncles, etc. I had blood samples from over 1 000 people.

*TP: You went to people’s homes and took blood samples, didn’t you?*

AH: Oh yes, I was known as the Vampire of the Fredrikstad region. My wife says that when children saw me they would cry and run. It was not that dramatic really. But there was no hint of informed consent or anything of that sort. The doctor said, “we need a blood sample”, and you did as requested. Very few people refused.

*TP: When you say markers and linkage -- to find the chromosomal locus for the disorder – you mean proteins, right?*

AH: Yes, but I’ll tell you about the DNA first. This is a humbling story. I carefully separated and froze plasma, serum and red blood cells. I threw away the little thing in the separation phase.

*TP: The buffy coat with the leukocytes?*

AH: Yes, the buffy coat with the DNA. I started collecting samples in 1970 and DNA was not on the agenda. In 1974, there were two guys called Goldstein and Brown who talked about their work at meetings -- they received the Nobel Prize in Physiology or Medicine in 1985. They did groundbreaking work on the lipoprotein receptor -- cell studies -- and proved that the receptor was deficient in familial hypercholesterolemia. This was when I was finishing my PhD thesis. I understood rather late in the game what I should have done – by then the DNA was in the garbage and everything else was frozen.

*TP: What did you find?*

AH: I think I proved that some patients lived rather longer than expected. This was pre-treatment days, way before statins. At most, you could prescribe diet. Compliance was so-so, understandably. Cholesterol in those days was much higher in Norwegians than it is today. With the lack of receptor for the lipoprotein, you would have a rather early demise – but so did everyone else. Some people survived fairly long. My oldest patient who had a tremendously high cholesterol, around 20 millimoles per liter, was 96. The disorder was very heterogeneous clinically. With the British researcher Joan Slack, we showed that family history was probably as good a predictor of early death as cholesterol level, indicating a role for genetic modifiers.

*TP: You did your thesis on hypercholesterolemia and qualified as a medical geneticist simultaneously?*

AH: Yes, by present day standards we came relatively easy to qualifying as specialists in Medical Genetics. We provided genetic counseling for some families, but the caseload was not that heavy. It’s much more demanding today. I continued with counseling along with the lipoprotein work, which is a bit paradoxical when I think of the teaching we received in genetics as students. Jan Mohr taught us about Mendel’s laws, illustrated with experiments with fruit flies. Genetics seemed not very relevant to clinical practice. It was a bit of a paradox that three of us, including Helge Boman, Harald Torsvik and myself in a class of 100 students became medical geneticists.

*TP: An example of how random events figure into your story.*

AH: Yes, I was primarily looking to do a PhD. Gradually I became more fascinated by genetics. I went back to Internal Medicine for a year after I completed my thesis. Then I got an offer from Kåre Berg who had arranged for the city of Oslo to fund a medical geneticist in the Department of Public Health -- to raise awareness and provide counseling.

*TP: When would this have been?*

AH: 1976. Since then I have largely worked in Medical Genetics with rare disorders as a unifying theme.

*TP: What did you actually do in 1976?*

AH: Genetic counseling. I had an office in one of the old barracks on the University of Oslo campus. A large office, but with very little equipment – myself, a bench and a telephone. The building I was in was supposed to have been torn down 30 years earlier, after the Second World War; it was definitely past its prime.

*TP: Who were your colleagues?*

AH: Kåre Berg and Carl Birger van der Hagen who did cytogenetics.

*TP: Carl Birger van der Hagen is the father of cytogenetics in Norway.*

AH: Yes. There was also a woman with a science background, Marit Hornberg Solaas. There were other people around as well. Helge Boman was there when I started in 1969. He subsequently left for Seattle to study with Arno Motulsky. Upon returning to Norway, he started the Medical Genetics Department in Bergen. Tobias Gjedde-Dahl jr., also a prominent figure in Medical Genetics in Norway, had gone to Tromsø. Karen Helene Ørstavik, who became very important later, hadn’t started yet.

*TP: Who came to see you in the mid and late 1970’s?*

AH: We held a course for the public health nurses who were very important because they saw families in the well-child clinics. The physicians in these clinics changed frequently and only worked part time. The nurses provided continuity. They became aware that genetics could be important and started to refer families. I remember some families very well. For example, a Pakistani family with a child with a Meckel-like syndrome that I have contact with still. Although I wouldn’t call the genetic counseling successful. The parents were double first cousins, and in subsequent generations, double first cousins are still marrying each other, at best proving that the counseling was non-directive

*TP: You must have seen some families with children with cystic fibrosis.*

AH: I did. I soon realized that I needed to make my way into clinical departments. There were three pediatric departments in Oslo. At the smallest one at Aker Hospital, a municipal hospital, I developed a good relationship with the pediatrician who cared for children with cystic fibrosis, Øystein Aagenæs. Then an important change came about. Karen Helene Ørstavik joined the department and we became involved in a summer camp for children with hemophilia.

*TP: When would this have been?*

AH: 1972 or thereabouts.

*TP: I should ask you about Esther Sanengen now.*

AH: Yes, Esther Sanengen was a great woman -- the founding mother of all work for families with rare disorders in Norway.

*TP: What was her connection to hemophilia?*

AH: She was recruited by two important people in Norwegian medicine, Peter Hjort and Christian Borchgrevink, to do something for boys with hemophilia. Because she was a well-connected woman with an iron will, she managed to put together what, at the time, was called an “Institute for Hemophiliacs”.

*TP: Why did hemophilia blaze the way for care for families with a rare disorder?*

AH: Well, there was the same kind of evolution in Norway as in most European countries. It started with hemophilia. Subsequently, a long period where nothing happened followed. Then various groups, like families with children with cystic fibrosis, formed organizations.

*TP: Was it because families with children with hemophilia organized themselves that something started to happen and that Esther Sanengen was conscripted?*

AH: Yes, that’s a good general description. Resourceful women saw the need families had for meeting other families and sharing experiences.

*TP: And what did Esther Sanengen do for them?*

AH: A social worker originally, she formed a group that went to the Department of Health demanding that something be done. She was met by a patronizing attitude, “go home a do a charity bazaar, dear lady”. She became angry and mobilized several of her husband’s important friends, and secured funding. This was an unheard in Norway; it’s “the American way”. We don’t do fundraising -- we expect public funding. She established a small facility where boys with hemophilia and their families could come. With time, she managed to expand the program and establish public funding. She had important contacts, influential physicians, with whom she worked closely and was able to make her voice heard.

*TP: The summer camps where boys with bleeding disorders could meet and have fun were one venture, but wasn’t Esther Sanengen also instrumental in initiating home treatment?*

AH: Yes, but that came later. This was early days. I remember as a clinical lecturer in Internal Medicine I took groups of medical students to the Hematology ward to meet patients who were on their backs for weeks because of joint bleeds, psoas hemorrhages or other terrible complications. And then when they were slowly mobilized, they would bleed again -- readily available targets for educational purposes.

*TP: Back to what you did at the summer camps.*

AH: Karen Helene Ørstavik and I provided genetic counseling. This must have been around 1972.

*TP: This was before molecular genetic testing was available. Factor levels were not reliable for carrier testing in females. You were doing Bayesian risk calculations, correct?*

AH: Yes. Karen Helene has a mathematical mind and was good at this. We addressed issues like “how many unaffected brothers could you have and still have a significant probability of being a carrier?” Rather primitive counseling compared with the present day.

*TP: Tell me more about working with the pediatric departments in Oslo in the beginning.*

AH: Well, I saw many children with malformations. These were the early days, “pre-syndrome days”, if I may use that expression. We were unable to recognize almost any of the syndromes that have been characterized today. We probably missed a long list of diagnoses that would have been made today.

I also started to work with social workers and other professionals who cared for families with disabled children. This taught me much about the burden of having a child with a handicap. I started seeing patients with physical therapists, social workers and nurses – people who knew the families and who could follow-up better than I could after a single genetic counseling session.

*TP: Therein lays an acknowledgement of the fact that life mostly takes place outside the consulting room.*

AH: That was an important learning for me.

*TP: Let’s go back to genetic counseling for a moment. If there is a key insight you could magically transfer into the heads of young colleagues, what would it be?*

AH: I think the most important thing is to listen to the patient’s agenda. I approach a consultation with my agenda – which is not necessarily similar to the patient’s or the family’s – and not necessarily relevant in light of their concerns and beliefs.

*TP: But do people always know what their agenda is?*

AH: No, I start by asking what the issues are and they come up with something. Gradually – it’s a dance of sorts, backwards and forwards – I try to elicit their concerns and broaden the scope of the conversation. Mostly I listen and ask open-ended questions. It can be difficult because at the back of my mind I have my agenda.

The internet has helped most families a lot. People often have some background information and a better idea of what they want to know. In the old days, the doctor was the sole source of knowledge. In one sense, counseling has become more demanding. On the other hand, it is often easier to get people to talk. However, the internet is also a source of misinformation. People sometimes have ideas lodged in the backs of their minds that they would be better off without.

I am also concerned about an information gap. By this, I mean that we are probably not seeing all the families and patients we should see. There is a pretty broad base for referrals from Pediatrics, etc…but some families we do not see and do not care for properly – for example families with minority backgrounds -- these families are unaware of the potential value genetic counseling could have for them.

*TP: Now you’re referring to the relatively recent influx of non-European, non-North American immigrants to Norway, correct?*

AH: Yes. From my experience working with minorities we encounter beliefs, for example about “the evil eye” and the like, which we do not address properly and which require more familiarity with the family’s background.

*TP: Back to your work history. What next?*

AH: To Frambu, a center for rare disorders about a half an hour by car from Oslo. I was there from 1985 to 1995.

*TP: What was Frambu when you started there and how has it evolved?*

AH: It started after the Second World War and the polio epidemics in the early 1950’s. Frambu was founded as a kind of holiday center for members of the youth movement in the Labour party; it’s been completely apolitical for many years now. Gradually, it evolved into a facility with summer camps for children with handicaps. The mothers started to talk among themselves when they dropped off and picked up their children. The children had various problems: intellectual disability, epilepsy, physical handicaps, spina bifida – they were children who were unable to attend regular summer camps. Their parents felt that it was safe for them at Frambu. There were doctors on staff, but no elaborate medical care. Gradually courses for mothers and children were established. Eventually, some fathers asked why they were excluded. Next sibs were included. But still, the focus was on relatively common disorders and disabilities.

Esther Sanengen who we talked about previously, arranged for boys with hemophilia to be able to attend the summer camps. This transitioned into courses for families. Cystic fibrosis followed as the next rare disorder.

In 1978, a woman who became very important in the rare disorder field, Lisbeth Myhre, who had osteogenesis imperfecta herself, wanted to start an advocacy organization. In the old days, we had established a registry for inherited disorders. It was originally funded by the Rockefeller foundation. We had permission – by the standards of the day – to collect data and had a list of people who had osteogenesis imperfecta. We wrote to orthopedic and surgical departments asking if they knew of more affected individuals. With the help of Esther Sanengen we organized the first course for osteogenesis imperfecta at Frambu in 1979. A big success. Over 50 of approximately 250 people in Norway with osteogenesis imperfecta attended. Everyone learned a lot: affected individuals and professionals alike. Orthopedic surgeons attended. We saw all degrees of severity and saw the consequences of prolonged immobilization. At this time, the transition to using metal rods and plates was happening. Previously, treatment consisted of lying on one’s back for several months waiting for fractures to heal. Some individuals had had more than 100 fractures. Most had not met anyone else with osteogenesis imperfecta before.

That was the start. From there we organized other groups through the Department of Medical Genetics and my personal contacts. We had a course for Huntington disease – affected individuals, children, partners, at-risk relatives – altogether about 75 people. Dynamite -- we saw people with Huntington disease at various stages and could really observe the disease in all its aspects. Next, we worked with individuals and families with neurofibromatoses and other rarer syndromes -- Williams syndrome, Prader-Willi syndrome, tuberous sclerosis, Cri du chat syndrome. Frambu helped the patients and their families start advocacy organizations for approximately 30 rare disorders between 1979 and 1995 – so Norway was a European pioneer in this area.

We brought families with members with the same disorder together and provided a comprehensive medical course – we emphasized empowerment and psychosocial issues. Frambu had a large educational department that provided school and daycare for the children during the courses. Medical people tend to forget that much of a child’s day is spent in daycare or school.

*TP: You were at Frambu for 10 years from 1985.*

AH: Yes, very formative years for me personally and for the field of rare disorders as well. I learned about the implications of genetic disorders in a new way. My family also became involved. Our two daughters were recruited as leaders at the summer camps, and my psychiatrist wife led groups for relatives of Huntington disease patients

*TP: In what ways were your years at Frambu important?*

AH: Many examples. I was recently at conferences for the neurofibromatoses and for Huntington disease where I met people from lay organizations whom I have known for 20 – 30 years. I have been in the field a long time and it can therefore get a bit repetitive, but it is still important to listen to the patient’s perspective, understand her / his concerns. For instance, individuals with Huntington disease are concerned for their children and grandchildren, not so much for themselves, even quite late in the disease.

*TP: You mentioned that Lisbeth Myhre was important.*

AH: Yes, she became the head of a Norwegian umbrella organization for people with disabilities and from there she moved into government, the Department of Health, then the Directorate for Health and Social Services. She instituted a designated program for rare disorders and was very influential. Through her own experience, she was very aware of what was needed. The Norwegian government has contributed rather generously so that the lay organizations for rare disorders have been able to continue their efforts. The organizations were usually started at Frambu during the initial course. People got together, were encouraged to organize and received some financial support. The Norwegian government has consistently co-funded meetings for lay organizations for rare disorders.

*TP: Lisbeth Myhre was an important advocate for paying attention to life outside the hospital and clinic. She rebelled against the tendency to reduce people’s lives to their medical issues.*

AH: Yes. I think that for many rare disorders we must acknowledge the current limitations of what medicine can do today.

*TP: From Frambu you went to Rikshospitalet in Oslo. Did a position come up?*

AH: Yes, it was odd that the most highly specialized tertiary care hospital in Norway didn’t have a Department of Genetics in 1995. Patients and families were badly underserved in that respect. Especially given the explosion of knowledge, hiring a geneticist was overdue. Pediatricians were knowledgeable, but their perspective was different.

*TP: You were recruited?*

AH: Yes. I worked alone to begin with, and then acquired a secretary. I started as an adjunct to the Center for Rare Disorders at Rikshospitalet, which had grown out of the work with Huntington disease at Frambu. We were first housed in the loft of the Dermatology Department and subsequently moved to Pediatrics. I worked closely with the pediatricians. We recruited Karen Helene Ørstavik from the other big hospital in Oslo, Ullevål Hospital. As in the Old Testament, I had to wait for her for some years, but in the end, she came. Subsequently, Rikshospitalet and Ullevål Hospital merged and became part of Oslo University Hospital. We relocated from the Pediatric outpatient clinic to more spacious facilities in a neighboring building in about 2002 where Clinical Genetics is located today.

*TP: One of your major achievements was introducing genetics into other medical specialties at what is now Oslo University Hospital – neurology, pediatrics of course, but also neurosurgery, orthopedics, endocrinology, obstetrics and gynecology. How did you do that?*

AH: By walking around and making myself familiar, available. Looking for people with common interests. It was a gradual process. I think one of my strengths is a non-threatening presence. I don’t seem to be out to take over things from others. Gradually genetics made itself useful. One of the distinct advantages of Medical Genetics is that in contrast to other specialties, much of what you learn in medical school remains useful. You are exposed to so much of medicine by being a medical geneticist.

*TP: You are known for playing well with others. As a consequence, you were for example instrumental in starting a multi-specialist team for craniofacial disorders at Rikshospitalet / Oslo University Hospital. This was one of the first multi-specialist teams in Norway.*

AH: We started more or less as a cleft lip and palate team and expanded from there. I remember giving a presentation at the big weekly meeting at Rikshospitalet for all the staff and showing videos of major craniofacial surgery. The goal was to present what these surgeries, which could last up to 12 hours, were really like without anyone in the audience fainting. The people who do these surgeries are impressive and you would never meet them if you confined yourself to your office.

From the patient’s perspective, it may be overwhelming meeting many doctors at the same time in a multi-specialist consultation. However, patients also benefit from this type of coordinated and effective management where common treatment goals are established – which operation first and why?

*TP: Another element of your success in bringing genetics into the mainstream of clinical medicine is that you show up. You’re dependable.*

AH: I’m not a good judge of myself but I think I’ve always been pretty dependable. I’ve tried to be there when there is a need – and I think people have had confidence in me because of this. And it’s has been rewarding – I have felt appreciated, too. I have good relationships with most people and seldom make enemies. I don’t like quarreling, but will if necessary stand up for my views.

*TP: Let’s talk about teaching, which you like. Which teaching have you enjoyed the most?*

AH: That brings me back to the lay organizations. I think I have learned most from teaching groups of parents of children with various disorders. You are asked questions that you usually don’t get in clinic. In a group, people often raise more demanding and interesting questions. There’s a shift in power. You have to scrutinize your values and beliefs and can’t get away with easy answers. You have to be able to explain and reason. It’s not good enough to just make claims. That’s the kind of teaching and interaction I like best. Academic lectures are less challenging in a way.

*TP: Which brings us to your academic work. You’re Norway’s first professor of genetic counseling at the faculty of Social Sciences at the University of Bergen, is that correct?*

AH: Yes, I have long experience in genetic counseling and contributed when the first Masters program in genetic counseling for people with various backgrounds – nurses, social workers, etc.. was started in Norway. And there’s another discussion. Who should do what in genetic counseling given the explosion of knowledge? You need doctors and genetic counselors. I think the field is changing so fast that we can’t see how medical genetics will evolve. There is a huge amount of technical data – there is a need for people with a counseling background, people with a medical background and people with a strong science background.

*TP: I could see increasing mainstreaming with genetic counselors and specialized nurses doing a lot of medically speaking straightforward counseling – which could be complicated enough in other respects. Medical geneticists could come to work more and more in teams with other clinicians taking on the medically complicated cases.*

AH: Obviously, there is task shifting but even so-called simple genetic counseling can be very complicated. For example, explaining autosomal dominant inheritance in Huntington disease is straightforward. However, determining whether people are mildly affected or not, and addressing some medical issues can be quite complicated. I think, therefore, genetic counselors need a repertoire of disorders, which they know really well -- including knowing all there is to know about medical issues.

*TP: You have served on many committees. Your advice has been sought in many contexts, for example, by different branches of government. You were also the leader of a regional ethics committee, the Norwegian version of an institutional review board, for six and a half years from 2008. Do you think such committees work well when assessing projects that have to do with genetics – do they have the necessary knowledge and skills?*

AH: Complicated question. Instead of obstructing research, I think we need to think about ethical aspects of research. It’s a bit like Clinical Genetics in the sense that the days of paternalism are gone. You need to advocate for patients’ interests. I think a part of medical ethics is insuring a minimum standard of informed consent. In our committee, we reviewed about 20 – 25 applications 10 times per year. Most projects were approved quickly. The committees are not a major hindrance for researchers. Our presence forced people to think about ethical aspects of their projects.

The second part of your question is more complicated. I recently attended a conference in Vienna on biobanks where it was asked whether the informed consent form is outdated because of all the new knowledge available via biobanks. Everyone could be sequenced. What should we do with all the information? Is it possible to give people information about all the technological possibilities, about the implications of having all their genetic data? There is no clear answer.

The dynamic informed consent is much talked about – a form for consent where people are kept updated via a website and may be asked for to give consent sequentially for new research projects. A one-time consent isn’t valid in this time of evolving research. All old informed consents become invalid in a way, especially for valuable large population-based longitudinal cohort studies like the Norwegian HUNT study, which is now starting a fourth data collection. Consent is a headache for everyone involved. There are few “retractors” among Norwegians, very few opt out. However, that may be because people are not sufficiently aware. At present, this issue is evolving and no one has the answer. Today, Norwegians seem to be willing to take some risks in order to contribute to research.

*TP: Let’s hope that researchers don’t abuse the trust they have been shown.*

AH: There are mostly theoretical concerns at this point. We have few examples of unfortunate events. However, the history of medical research is full of incidents that we are not proud of – and they can’t always be excused because we didn’t know better. Famously, Gerhard Armauer Hansen from Bergen was convicted for performing unethical research on a girl with leprosy. That goes 145 years back. He was sentenced and lost his position because of unethical conduct.

TP: Again talking about the big picture. Are you worried about the potential for misuse of some of the new technologies? Does it keep you awake at night?

AH: No. I think I am pretty optimistic and non-ruminating about things in general. We have been hyping personalized medicine for a while, but I think there is a future for medicine that is better tailored to the individual. I think with increased understanding of genetic mechanisms we will be able to target much more precisely the patients who will profit from certain treatments. The trend is evident in oncology, spinal muscular atrophy, Duchenne muscular dystrophy and cystic fibrosis, to mention some current examples.

TP: The poster child for personalized medicine is rare disorders and especially rare metabolic disorders that can be treated effectively.

AH: But interesting things are happening in other areas as well. Take Huntington disease – among the most devastating of all the incurable disorders. Antisense nucleotides seem to hold some promise. But new therapies can be prohibitively expensive. Enzyme replacement therapy for the mucopolysaccharidoses is very expensive even for a rich country like Norway. Apart from orphan drug legislation there is little incentive for Big Pharma to develop new drugs.

TP: New topic. You’re interested in history. But we’ll limit you to the history of genetics. If you were to invite three geneticists, living or dead, to a dinner party, who would they be?

AH: Interesting. I’ve had the chance to meet several great individuals in medicine. I met Victor McKusick and even get to touch Osler’s desk, which remains in McKusick’s department. I would invite Victor McKusick. He was a great man who contributed so much to the organization of knowledge. I would also invite his wife Ann, a delightful person. The next guest would probably be Peter Harper. He made a big impression on me. He trained with Victor McKusick. I spent a few days with Peter Harper and his social worker, Audrey Taylor, early on in my 40-year long career with Huntington disease. From them I learned some of the basics of working with individuals with the disease and their families. I would also like to invite two, almost honorary Norwegians, namely Dian Donnai from Manchester and Han Brunner from Nijmegen. They’ve come regularly to Norway for years to teach dysmorphology, basic molecular mechanisms and how to integrate new knowledge into the clinic in a useful way. I know I have invited more than three, but Di and Han are linked together for me.

TP: As for the future, do you think that Medical Genetics is a good specialty for a young physician to choose? Are we entering or exiting the golden age?

AH: Definitely entering a golden age. As I said earlier, I think clinical geneticists can find a use for everything they learned in medical school. But we need to learn more basic science. It’s a broad specialty; no one can master the entire field. Whether we are splitting and for instance dividing ourselves into clinical geneticists and medical geneticists is debatable. I still think there is a need for clinical skills as well as working in the lab. I think we have many years of the golden age ahead of us even though parts of what we do can be automated. We can for example take a picture of a face and ask Dr. Google for a diagnosis.

TP: Isn’t artificial intelligence just another tool?

AH: Yes. I’m optimistic with respect to the future. There is definitely a need for more rapid diagnosis. That’s changing. New sequencing technology is changing things fast. But still you need someone to pull the bits together and explain.

TP: In Norway you can go straight into Medical Genetics after you get your MD and completing a 1 ½ year of training whereas in other countries you have to do a substantial amount of Internal Medicine or Pediatrics first.

AH: I think you definitely need some years of clinical training. I’ve found it very useful. I think you need some Pediatrics to be a good clinical geneticist.

TP: For someone who has been so-called retired for nine years, you are still pretty busy.

AH: I think I spend more than a full workweek on different issues related to Medical Genetics. I work differently. I’m still interested in certain disorders -- the neurofibromatoses, Huntington disease and the mucopolysaccharidoses. I give lectures on, and have recently attended European conferences about, these disorders. I think you need something to keep your brain going. I also have a very active wife who works politically on issues for the elderly. We sometimes sit down for formal planning meetings. I’m busy and enjoying myself.

TP: And you can choose what you want to do.

AH: Yes. I’m a member of the National Board of Medical Ethics but that is a small task compared to heading a Regional Ethics Committee. The caseload is much less but the debates are interesting.

TP: I think of you as a person who has put a lot of time and energy into simply trying to help people.

AH: Thank you for that.

TP: Do you have regrets looking back?

AH: There are decisions which could have been different, of course. Personally speaking, like all fathers my age, I could have been a better father, been more present in my children’s childhoods. But otherwise, I am pretty satisfied with life.