Journal of Clinical Immunology (2024) 44:40
https://doi.org/10.1007/s10875-023-01645-x

"The Regimental Pediatrician": Driving in the Wilds

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Published online: 13 January 2024

Columbus' Egg

As the years went on, I started to wonder more and more about whether I could manage to be transferred from Boden to more southern climes. I had applied for two posts but both times I had been out-distanced by a fellow applicant with a PhD in medicine. Would my colleagues in Stockholm's pessimistic predictions about my future turn out to be right?

I had four sons, who were going to need an education. There was a secondary school in Luleå but no opportunities for higher education closer than Uppsala. I understood that I probably needed to improve my qualifications by producing some sort of academic work. But what on? What subject? One couldn't "write a book" about a collection of individual, rare cases. I had had a number of cases involving ECG changes in babies and small children which I had interpreted as indicating viral myocarditis. But that was hardly enough material. Furthermore, I found it difficult to interpret changes in ECG results as I had only studied elements of this specialist area from books, and the field had quickly grown bigger and become more sophisticated. And I had no university clinic, no professor who I knew who might support me.

Then—exactly when I most needed it—my usual luck came to my rescue.

In March 1949, a two-month-old baby girl from Överkalix was brought to the hospital. She had a high temperature and otitis with mastoiditis. Also, in several places, she had bean-sized and larger septic-looking boils on her skin. Boils, "spots" on the skin, are almost never seen in babies. Blood samples showed that the baby had total agranulocytosis! I had never read of or heard of agranulocytosis in infants. What on earth was this? Had the baby been poisoned in some way? Had she been given any medicine? No.

Further questioning of the mother revealed an astonishing fact. This was her ninth child and four of her previous children had died in infancy with high fever. Three of them had had boils like this one! When the mother was also able to say that similar cases had been seen in babies in the father's family, I did not need to be a genius to work out that I was on the track of a previously unknown, hereditary disease. On top of that, a hereditary, disastrous granulocytopoesis problem. There was, I knew, no description of any hereditary disruption to this blood cell system—apart from what was known as the Pelger-Huet anomaly, which is not a disease.

It struck me, like lightning, that here—*here*—I had a subject to write about. If I could manage to bring the thing off, if I could collect several such cases, living or dead, and map their family relationships, I would have to behave in an unusually asinine way not to be able to manage to produce a thesis at least worthy of a pass mark.

I started by borrowing a car and driving up to the parish office in Överkalix. It soon emerged that one of the related children had died in Gällivare hospital. It had had boils and the diagnosis read agranulocytosis! (And the blood samples, bone marrow samples and samples for pathological-anatomical diagnosis had been preserved!) Victory, victory—beyond belief!!

I spent two days up there searching through the church registers. Using these books correctly proved to be a difficult task. I did not yet know the technique for finding what I wanted to know in the church archives. Genealogy is a skill that needs to be learned. On the second day, the parish priest told his assistant to help me. This was a very serious, pale, thin young man. I asked him if he could find me information about the families of the father and the mother, when he had the time. I needed the names and dates of birth of parents and children. And any death dates for babies and small children. And any diagnoses. His face filled with vertical frown lines, he undertook the task.

It was more than a month before I heard from him. During that period, I was treating the "key case". The girl was given penicillin and the boils healed. However, the agranulocytosis remained. And soon new boils appeared. And then one day, I received a

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large, thick letter from the parish office. It contained about ten typewritten pages packed with data. The assistant had completed his mission—and in much more detail than I had asked him to. He had traced the ancestry back and provided accounts of a number of old families that had died out. It was interesting and intriguing to find families with 12 to 15 children, 5 or 6 of whom had died as babies. For reasons unknown—of course. And there were no living people to ask. On the basis of this information and with the additional data, I subsequently obtained myself; however, I was able to calculate that infant mortality in the "family" was 4–5 times higher than in the district in general!

But the account of the families that were still living was what interested me the most. And there I made a discovery! Lying on my stomach on the dining room floor, I drew the members of the family and the relationships between them on a big sheet of brown paper. It was soon clear that the parents of the key case were third cousins. And that their siblings had formed families and had several children, among whom "obscure" deaths in infancy had occurred.

New journeys to Överkalix by car, this time to talk to the provincial doctor and his district nurses and midwives. The doctor, who had not been there long, knew nothing. But the interviews with some of the older district nurses and midwives were rewarding. In actual fact, they already knew there was a mysterious, fatal illness involving boils among babies—not just in the Överkalix district but in the Gällivare area too. They had even given the illness a name. They called it "Diirivara boils". Diirivara was the village the key case came from!

At an early stage, I realised that the disease was recessive. The parents of the ill children were always healthy. Blood samples from them and the healthy children were normal. It soon also became plain to me that it was a question of what was termed a lethal gene. A deadly characteristic. The people affected were defenceless against bacterial infections. All of them died. Most died before reaching the age of two. Antibiotics of various kinds proved capable of extending their lives for a few more years—in the most fortunate cases. But all died as children.

Luckily—for me—as well as an expression of the agranulocytosis of the cases, the symptom of boils was almost compulsory. I ought therefore to be able to work out retroactively which of the dead babies had had the disease—if the parents were living and I was able to question them. The death and funeral register held by the church was no use in this respect. The causes of death, the "diagnoses" written down there were lacking. Strikingly often they stated: "Born weak".

First, I thought that premature birth was a distinctive feature of the district. But it turned out that this was not at all the case. In a case where I had a definite "catamnesis", I was able to work out the course of events:

The baby was born one winter in a remote settlement with no road link. The midwife never reached it. Nor did the district nurse. After 2–3 weeks, the baby developed a fever and boils. The baby died after a week or two. The settlement was cut off and it was impossible to reach habitation. The dead infant had to lie in the woodshed for a week or so. Then the father was able to put the little body on a sled and pull it to the church and the priest on skis. The priest naturally asked what the little one had died of. And the father said—as was true—"He was weak". So the priest wrote "Born weak" in the big book! That was how it went, back then—and probably still does to this day sometimes.

There was a lot of travelling, and a lot of ploughing through church archives in most of Norrbotten's churches and also in the county archives in Härnösand. When my thesis was finally complete, I had a card index with about 700 lineages.

When the work was completed in 1975, I had 1700. It was fortunate that my topic happened to be a blood disease. I had been fascinated by blood and blood diseases since my student days, and as a medical student, I spent time on bone marrow diagnosis with reader Nils-Göran Nordensson in a post at Saint Erik's hospital in Stockholm. Pictures of bone marrow had fascinated me ever since.

But the fact that my subject required such inordinate effort in terms of genealogical research was truly *Nemesis Divina*. Few things interested me as little as how people were related to each other. When I started my work, I was barely able to work out what a cousin was without pen and paper! Since then, I have had much to learn. It was quite a baptism of fire.

Driving in the Wilds

Driving a car alone up there was sometimes a little nerve-wracking. I was not an experienced driver and populated areas were few and far between. The thought of the car breaking down or getting into some other kind of difficulty was not reassuring. On one trip, on a stormy autumn day, my journey came to an abrupt halt. The road was blocked by a fallen tree, and it was impossible to turn round. Luckily it was no more than 20 min before a car drove up behind mine. The driver leaped out with a cheerful shout. He was equipped with a saw and an axe. In a few minutes he had sawn the road clear and we continued on our way. He had to saw through two more trees before I arrived "home" in my guesthouse in Landsjärv. After that day, I never travelled without the necessary tools. In the winter, besides a saw and an axe, one always took a snow shovel and skis and, most importantly of all, a litre of alcohol (apart from the stuff one had already poured into the petrol tank). The garages were vast distances apart and 10 L of "soup" in a jerry can in the boot was essential kit.

Besides engine failure, fallen trees and other "natural disasters", there were more hazardous things to be encountered. It was not that uncommon to come up against lorries driven by barely sober drivers with their foot to the floor. They would appear coming round corners on two wheels—not particularly bothered about which side of the road they were on. It was a bit worrying and one would always keep an eye on what the terrain was like to the side of the road in case the safest option was to leave the road voluntarily.

There were many stories about drunk drivers up in the north. One ran as follows: Empty lorry. Driver, and driver's mate sitting beside him. Both drunk. They were taking the long, dead straight road through the marshes northwards towards Korpilombolo. First, the driver's mate fell asleep. Then the driver fell asleep. The car started to veer off the road. When the willows hit the windscreen,

At the end of the path

the fellow in the passenger seat woke up and shouted: "SECOND GEAR—THICK FOREST COMING!".

The story might be made up but the following apparently isn't:

In the middle of winter, an inebriated party was driving on the road from Kukkola (on the Torne river) towards Björkfors in the middle of the night. The car went off the road on a bend and spun round in a cloud of snow. The driver came to his senses—eventually—and saw in the moonlight that some of the passengers had been thrown out of the vehicle. All was silent and still and none of them was moving.

In the court room, the judge asked the driver what he had done next: "Run down the road—Leipijärvi—found cottage—banged on window—shouted—UP MAN!—IN THE FOREST!—CARRY CORPSES!—got back—corpses sitting up and eating sandwiches".

On the Trail

Naturally, the first thing I had to do was trace cases in living memory that had died as babies with symptoms of boils. I borrowed cars from friends in Boden and from the county council. But the roads that were passable by car did not always reach my destination. Sometimes, I had to travel on foot on paths through the undergrowth and on planks over bogs—for kilometres—to reach the house I sought. Sometimes it was hard to find where the path in question began on the map.

The people I visited fortunately all spoke Swedish—if they had to. As a rule, they were better at Finnish and their Swedish was not always easy to understand. The genuine, ancient Överkalix dialect, for example, was more impossible to understand than the Gotlandic they speak on Fårön. It sounded almost like English—except one couldn't understand a bit of it. However, they could also speak "proper Swedish"—but in a rather unique way. I have always had a poor ear for languages and I had to up my game.

These people—far out in bear country—rarely met people they didn't know. They were shy and suspicious. Saying they were taciturn would be putting it mildly. They were people of extremely few words, and most of their answers to my questions were monosyllabic—"aye…, ahh…, naaah…".

Asking questions directly, immediately stating one's errand and why one had sought out their cottage were impossible. One would probably not have been given any information at all. No, the job required different tactics—and very time-consuming ones. A visit often proceeded like this:

Of course you would have been observed from a kilometre away, balancing on the wooden slats, picking your way across the last bog. When you reached the cottage, you had to knock on the door several times before unclear noises from within indicated that you might enter—cap in hand.

Usually one drew a blank

The father would be sitting at the kitchen table, hidden behind a newspaper (a week or so old). Only the mother's back would be visible as she was stood by the wood-fired stove stirring a pan. Any children would have gone into hiding long ago.

"Good day, good day", you said. After a long silence a "good day" would be heard from the father, who lowered the newspaper for a moment before swiftly returning to his reading. The mother would now set a wooden chair in the middle of the floor. According to custom (which I had to learn), the stranger was then to take the chair and place it by the front door before sitting down. Now you could say who you were. After that, you started to talk about the weather, which had been terrible recently, hadn't it? How had the autumn harvest gone? Would the crops ripen in time this year? And so on, and so on. All the questions were answered reluctantly.

"Ah…", "Well…", "Indeed…" sometimes a bit more.

By now, the mother would have made coffee and set out the cups on the table. The father would put down the paper and coffee drinking commenced.

Now it was time to ask how the children were. The answers started to get slightly more detailed. After that—very carefully—you said that you knew that they had lost one or two babies. "Well… umm…. happen as we had." Now they started to grow more talkative. Talking about illnesses is a universally appreciated topic of conversation.

In that way, I could find out what I wanted to know.

Seeking out such a family and conducting the interview naturally took a whole day. Sometimes, my efforts were rewarded. Usually though, one drew a blank.

"The Book"

The first result of my work was an essay on the disease published in *Läkartidningen*. I also spoke on the subject one autumn at a national medical meeting in Stockholm. I thought I would complete my thesis in 1951 or 1952. But it was not to be. I fell ill.

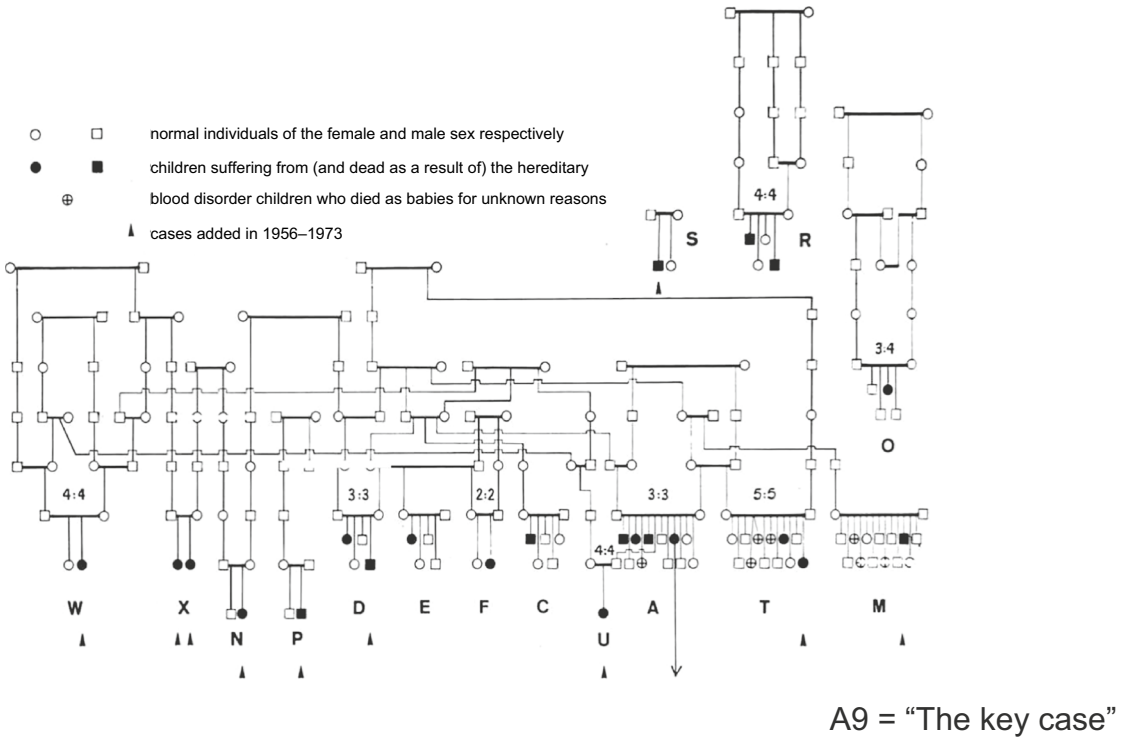
I had a severe episode of hereditary (endogenous) depression, an illness that relapsed and plagued my life with ghastly regularity. The fact that it all began in 1950 was naturally due to exogenous, external causes. For almost ten years, I had had a very large workload and had "burned my candle at both ends". Personally, I believed (as did everyone else) that the blame lay with my prospective thesis. But after 1956 when—in an "intermission"—I successfully defended my thesis, I did not become better in the slightest. The first ten years—before the modern thymoleptics became reasonably effective—were the hardest. Having researched a hereditary disease, I now got to experience for myself what inheriting "unpleasant" genes can mean. Fortunately, I had also inherited other, more positive and "nutritious" genes.

Although I had not completed my thesis at that point, in 1952, I secured a post in Norrköping. My practical work in Norrbotten was known. People were aware that I had a relatively unique thesis in progress. That was enough.

My thesis was not completed until 1956. During that period, more, valuable new cases were added to my material. A couple of these had been treated and undergone special investigation at the children's clinic in Uppsala. As my thesis was then able to be produced from the Uppsala clinic—and not from an unknown hospital up near the Arctic Circle—it was assured greater weight, and greater impact internationally speaking.

My thesis aroused attention and over the years, more than 30 similar cases have been published from different countries. There is still a new case in Norrbotten every two or every three years. Four cases have been discovered further south in Sweden. No familial relationships have been able to be traced between these and the Norrbotten cases. The gene appears everywhere—but is rare. However, in Norrbotten, it is found in concentrated form.

It is naturally original to have "invented" a previously unknown disease. However, 5–10, perhaps 100 similar cases (and just as many deaths from it) occur each year in the whole world. At the moment, the disease is of little importance. However, the blood researchers working theoretically and experimentally think that the disease is a "key disease" for the future understanding of the factors that determine the production of granulocytes in bone marrow; factors concerning the production of stem cells and substances that govern the granulocyte maturation process. THIS is a joy for me. The disease might have a greater importance than I primarily thought. In no country in the world other than Sweden can one expect a newly diagnosed case every two or three years. As a result, there is an opportunity for the riddle of granulocytopoeisis to be solved—in Sweden. I hope that this happens in my lifetime.



Family tree showing the family relationships between the parents of cases of hereditary agranulocytosis in children in Norbotten

Most of the families were able to be linked together in one consistent "tree". Three separate families are shown in the top right. Regarding two of these, the families R and O, genealogical research was able to ascertain blood connections between the parents. Regarding the family S, its roots could not be established. One must assume that, over the two centuries that the family history spans, there may have been cases of illegitimate children. Another factor is that a couple of churches, including the one in Pajala, burnt down, destroying all the archives. There are sure to be familial links joining these three families to the big group. The different families were identified as A, B, C, D etc., in the order in which they were discovered. During the course of the investigation, some families had to be excluded due to a lack of documentation in some respect.

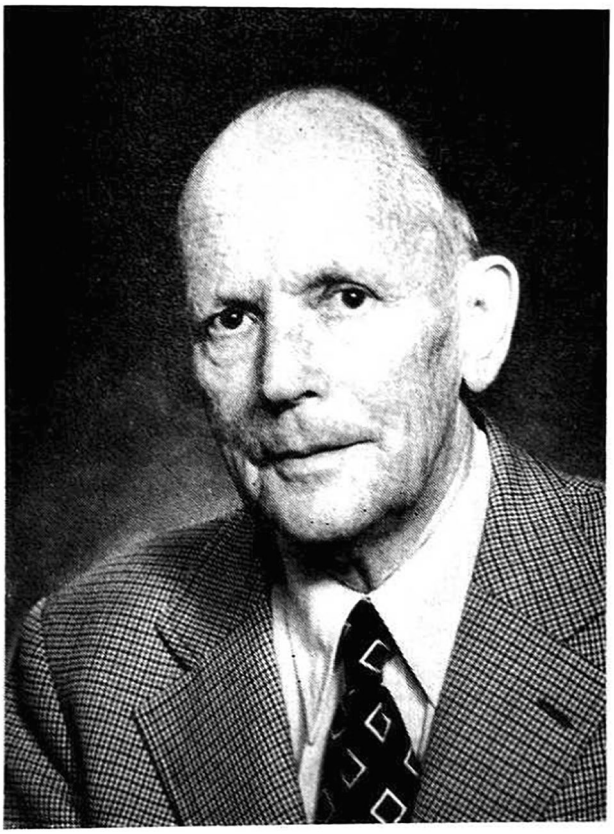
The designations 2:2, 3:3, 4:4, 5:5 denote that the parents were 1st, 2nd, 3rd and 4th cousins, respectively.

On the family tree, one can see the enormous amount of good fortune the author had in happening to encounter the ninth child in family A (A_9) as his first case. This was the "key case". Had I encountered, e.g. F_2 or N_2 or W_2 or S_1—I would never have realised that the disease was hereditary. Had it been the families X, D, T, R, it is possible that my detective instincts would have been stimulated but less likely.

In Conclusion

My father-in-law, who lived to a ripe old age, often said in the autumn of his years: "It is remarkable how well everything turned out for me". I would say the same. I have had luck and success throughout my life. The fact that Fate smote me with agonising periods of depression from time to time cannot, however, be categorised as luck. Nevertheless, they have been useful. I have learned to know myself and my limitations, and above all, I have gained a deeper understanding of the sufferings of others. I have fully appreciated that the gift of health is the most important thing of all. Furthermore—between the depressive episodes—I have been able to see and enjoy the beauty of nature and the multi-faceted richness of life in a particularly intense and clear-eyed way and—despite everything—have been able to gain more enjoyment than most.

It is utterly plain that my ten years up in Norrbotten were the high point of my life. Few people are granted the chance to perform such an interesting, exciting and rewarding job at a young age. Living in the present with every fibre of one's being and greeting every day as a new adventure.



Rolf Kostmann, 1979.

Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/s10875-023-01645-x.

Author Contribution RK wrote the memoir.

Funding Not applicable.

Data Availability Not applicable.

Declarations

Ethics Approval Not applicable.

Consent to Participate Not applicable.

Consent for Publication Not applicable.

Competing Interests The authors declare no competing interests.