

History of Rett Syndrome

In 1954, Dr. Andreas Rett, a pediatrician in Vienna, Austria, first noticed two girls as they sat in his waiting room with their mothers. He observed these children making the same repetitive hand-washing motions. Curious, he compared their clinical and developmental histories and discovered they were very similar.

Dr. Rett checked with his nurse and learned that he had six other girls with similar behavior in his practice. Surely, he thought, all these girls must have the same disorder. Not content with studying his own patients, Dr. Rett made a film of these girls and traveled throughout Europe seeking other children with these symptoms.

Meanwhile, in 1960, young female patients in Sweden with quite similar symptoms caught the eye of their own physician, Dr. Bengt Hagberg. Dr. Hagberg collected the records of these girls and put them aside, intending to return to them when he had more time to study this curious phenomenon.

Then, in 1966, Dr. Rett published his findings in several German medical journals, which, however well-known in that part of the world, were hardly mainstream reading for much of the rest of the world's medical community. Even after Dr. Rett published a description of the disease in English in 1977, Rett syndrome remained in the backwaters of medical concern: The pre-internet world lacked the electronic information highways taken for granted in the 21st Century.

But in 1983 an article on Rett syndrome appeared in the mainstream, English-language journal, *Annals of Neurology*. Written by none other than Dr. Hagberg and his colleagues, the report finally raised the profile of Rett syndrome and put it on the radar screen of many more investigators. This article was a breakthrough in communicating details of the disease to a wide audience, and the authors honored its pioneering researcher by naming it Rett syndrome.

As investigators continued to chip away at the shell of mystery surrounding Rett syndrome, increased research funding ensured that the work would continue. A team of scientists from Baylor University (Houston, TX) and Stanford University (Palo Alto, CA), toiled in the labs and clinics trying to pinpoint the cause of Rett syndrome.

A major breakthrough occurred in 1999, when a research fellow at Baylor named Ruthie Amir discovered MECP2, the gene that, when mutated, causes Rett syndrome. The discovery of the gene, located at the Xq28 site on the X chromosome was a triumph for the Baylor team, led by Huda Y. Zoghbi, MD, a professor in the departments of pediatrics, neurology, neuroscience, and molecular human genetics at the Howard Hughes Medical Institute.

The discovery that MECP2 is on the X chromosome proved that Rett syndrome is an X-linked disorder. And because only one of the two X chromosomes need have the mutation in order for it to cause the disorder, this is a dominant disorder as well. The fact that Rett syndrome is an X-linked dominant disorder also helps explain why it is usually found only in girls.

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by Kathy Hunter

Dr. Andreas Rett died at the age of 73 in his hometown of Vienna, Austria on April 25, 1997.

Our lives will never be the same for knowing him. Through his insight, dedication and compassion, Dr. Rett reached out to families all over the world.

He gave us hope and wisdom, and taught us to believe in our girls, to search their eyes for sparks of understanding that were invisible to the rest of the world.

He encouraged his medical colleagues to work together with parents as a team, to work with one another as a force for discovery.

Although very ill for the last several years, Dr. Rett never gave up on his dream to find answers to the puzzling disorder that took his family name.

Dr. Rett was one of those people that you may never have had the privilege to meet, yet who has changed your life forever. First he held our hands, and soon, he held our hearts.

Dr. Rett's work began over fifty years ago, after serving as a medic in a German navy hospital. After the war, he studied medicine in Innsbruck and Bonn and in 1948, went to Vienna. Realizing the great need for a children's clinic, he approached the mayor and other local politicians, who were very difficult to persuade in a community where disabled children were considered social outcasts. His patience was finally rewarded when the 100 bed hospital, Rosenhugel (Rose Hill) opened its doors to patients from all over Europe. It was the first facility of its kind in the world, and he was the only physician in Austria caring for disabled children.

It was a 70 year old building belonging to an institution for aged people with very few doctors and nurses, and no water faucets in the sick bays. Patients in wheelchairs had to be hand carried down the many flights of steps.

In time, Dr. Rett developed the clinic into a center for the handicapped, and with research funds he solicited, was able to provide adequate staff, the first EEG and the first clinical psychologist in a state hospital in Austria.

Dr. Rett first recognized Rett syndrome in 1954. He wrote a paper and made a film of several girls, which he took all over Europe, as he tried to convince others that the girls represented a distinct clinical entity. Very few listened, and those who did listen, did not believe.

He published several articles in obscure German language journals and continued his campaign. It was not until the disorder was reported in the English language by Dr. Bengt Hagberg of Sweden some 28 years later that Rett syndrome was finally recognized in 1982.

He often worked 16 hours a day, 7 days a week. When he took a rare vacation, he took to writing and during his lifetime published more than 300 medical articles.

He received numerous awards and distinctions in his career, but none which he prized more than the National Association for Retarded Citizens's 1989 Distinguished Research Award in which he was recognized as a pioneer in the care of mentally and physically handicapped children. At the time of his discovery of Rett syndrome, he was caring for over 5,000 handicapped people.

Always self-conscious about his command of English, Dr. Rett once said, "I never learned English, but I speak it."

Of our daughters, he said "they feel all the love given to them. They have a great sensitivity for love. I am sure of this. There are many mysteries, and one of them is the girls's eyes. I tell all the parents to look at their eyes. The eyes are talking to them. I am sure the girls understand everything, but they can do nothing with the information."

Of parents, Dr. Rett said, "It is the parents who make most of the diagnoses, not the doctors, who often tell mothers that they are hysterical. Such doctors are very wrong - they know nothing, but say something."

In a conference address, Dr. Rett shared his thoughts with parents and professionals. "These three words summarize best our task: To live, to love, and to learn. We are aware of the fact that many mysteries of this syndrome still remain undisclosed, and therefore, for the time being, we have no option but to live with it. However, the children with their very special ways give us enough impulse to share their lives. It is a further mystery of this syndrome that the affected children render it easy for us to love them. A dominating factor in the care of such children is that they understand us and we understand them. Their appearance and the sparkle in their eyes make it easy to love them. Daily care for them and working with them gives us grownups strength, enabling us to learn the special treatment required, thus furthering our own development. To live with them, to love them and to learn from them are the rudimentary principles of our work."

As I reminisce through the years and the many good memories, my eyes fill with tears. I remember his enthusiasm and passion, his strength and tenderness, his care, encouragement and guidance. But most of all, I remember his love. He called himself a simple pediatrician. We all know that he was a great man. We will never, ever be the same.

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