

THE RETT SYNDROME HANDBOOK

SECOND EDITION

BY KATHY HUNTER

Parent, Founder and President
International Rett Syndrome Association

*With contributions from the best Rett experts
around the globe and heartfelt stories
of love and courage
from fellow families everywhere*



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DEDICATION

To my daughter, Stacie, for giving my life purpose and inspiration ...

To my family for their continued patience, understanding and sacrifice ...

To the medical experts who give of themselves tirelessly and with determination to conquer RS ...

To the dedicated teachers and therapists who use loving hands and who recognize small wonders ...

To the contributors to this book, whose gifts of knowledge and experience light the way ...

*To Kim Poulos Lieberz, who always understands how to illustrate what I want to say,
and whose help with editing was an immeasurable gift of love ...*

*To the families in Rettland everywhere, who live ordinary lives yet meet extraordinary difficulties
with courage and conviction day in and day out ...*

*And to our loved ones with RS, who rise above the challenges every moment of their lives,
and through whose generous gifts of unconditional love
we find the hope and inspiration to go on ...*

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*Rett Syndrome
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16 Years of
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1989-2005*

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FOREWORD

How fortunate is the Rett community to have Kathy Hunter, founder and president of the International Rett Syndrome Association (IRSA), put together a new edition of the Rett Syndrome Handbook. This handbook will be useful to everyone who has been touched by Rett syndrome: parents, relatives, friends, neighbors, teachers, caregivers, physicians, and scientists.

Twenty years ago our knowledge about the child with Rett syndrome and her needs were very limited, but Kathy and parents like her shared their observations and experiences to inspire physicians and scientists to study this syndrome and to help other parents provide the best care for their children and cope with the challenges of this disorder. Today, there are over 1,230 medical and scientific publications on Rett syndrome. The amount of knowledge that a parent has to sift through or comprehend is overwhelming. This handbook introduces the reader to the nervous system and how it is affected in Rett syndrome, summarizes what medical research has informed us about this disorder and shares the best management options for some of the symptoms. The book provides practical advice about how to cope with the medical challenges and how to enjoy life with Rett syndrome.

It is written beautifully (as we have grown to expect from Kathy) and is easy to read. The book is rich with the experiences and the wisdom of many parents who want to share lessons they learned to help new generations of patients with Rett syndrome and families. The book also provides the reader with resources to find the necessary equipment, financial and community support, and up-to-date information. It concludes with updates on the latest research findings and ongoing research activities giving the reader a sense of what the future might hold. Kathy and IRSA have played a major role in increasing awareness about Rett syndrome, ensuring research support, providing families with up-to-date information, and sharing resources for support. On a personal note, I can say that their help went beyond advocacy and family support. I learned quite a bit about Rett syndrome from Kathy and the IRSA family, and their support helped us to identify the gene in 1999.

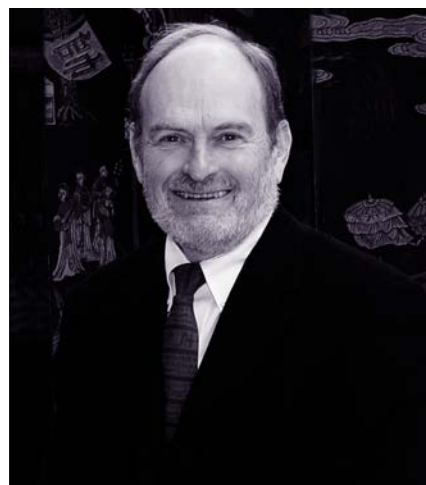
Watching all the exciting discoveries on Rett and the gene (*MECP2*), one can't help but wonder how much more this disease and its causative gene are going to teach us about the brain and the magic of its workings. The Rett Syndrome Handbook certainly captures all of this and leaves the reader wanting more. At the pace research is moving, we hope that more discoveries will come soon, and we can bet that Kathy will be happy to write a new edition to share exciting advances, and hopefully, effective treatments.



Huda Y. Zoghbi, M.D., Ph.D.
Investigator, Howard Hughes Medical Institute
Professor, Baylor College of Medicine



Who better to produce a comprehensive handbook on Rett syndrome than the Founder and President of the International Rett Syndrome Association? Kathy Hunter has more than thirty years of experience as a personal practitioner in the world of Rett syndrome, dedicated to providing the best and most current information for parents, extended family members, and other caretakers. This is a world that was never envisioned by her or any parent, but one that was faced directly in order to deal with the daily aspects of this relatively new neurodevelopmental disorder that affects predominantly girls and women. This handbook contains a wealth of information from persons, including physicians and scientists, therapists and educators, but of equal importance, parents of children with Rett syndrome, whose daily experiences surely qualify them as “experts.”



This handbook should serve as a valuable resource when needed. Not every aspect will pertain to every child, but virtually every aspect of Rett syndrome is covered in detail. The handbook is written with sensitivity and objectivity, but from the perspective that it is better to shed light on a subject than to curse the unknown. The Rett Syndrome Handbook is a treasure chest of useful and important information that should serve the reader well and advance the lives of those affected by this disorder.

Alan K. Percy, M.D.
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Director, Civitan-Sparks Clinic
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INTRODUCTION

In 1954, Dr. Andreas Rett of Vienna, Austria, noticed two girls sitting together with their mothers in his waiting room. The girls made the same unusual handwashing movements, and he discovered after examining them that their clinical and developmental histories were strikingly alike. After consulting with his nurse, Dr. Rett found six others like them in his practice and made a film of the girls, which he took all over Europe trying to find other cases. His findings were published in several obscure German language medical journals, which unfortunately never reached the attention of the worldwide medical community.

In 1960, Dr. Bengt Hagberg observed several girls who had similar behaviors in his busy practice in Sweden. He put these interesting cases in a special box under his desk, vowing to look further into the nature of this unusual and unknown disorder. In 1978, Dr. Ishikawa and colleagues from Japan described three girls in a brief note which also went unnoticed. Many years went by before Drs. Hagberg and Rett realized they were reporting the same disorder. In a generous gesture, Dr. Hagberg deferred his original descriptive title and submitted the name Rett syndrome for the first English language article on the disorder, published in late 1983. Until his death in 1996, Dr. Rett worked ceaselessly to unite parents and professionals in a community of care to bring a better life to the girls whose disorder bore his name. Dr. Hagberg, now in his eighties, continues today as a world leader in the field. His energy and enthusiasm in the research arena are only surpassed by his gentle spirit and compassion at the human level of the lives he works to improve.

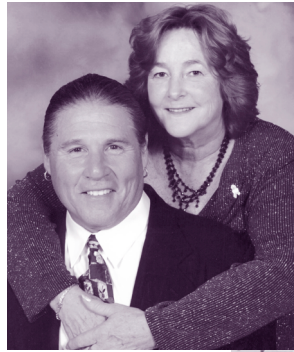
“I can only express my gratitude to all the parents for their love and the affection they give to their children, and my admiration for all their efforts and services they deliver to their girls. Don't lose heart in your work, keep your love for the children, and remember what I have always tried to say: watch the wonderful expression of the eyes of these girls, an expression which makes them so lovable.”

- PROFESSOR ANDREAS RETT



Shortly before the first paper on RS was published, my daughter, Stacie, who was ten years old at the time, was given the diagnosis of RS. She became the thirty-sixth documented case in the world. Stacie had developed normally until fifteen months of age, then began a regression that led to a loss of the few words she had developed, aloofness, withdrawal and irritability most of the time. She began mouthing and wringing her hands constantly. Over the years, and like many of your daughters, Stacie had a number of diagnoses, including autism and Angelman (Happy Puppet) syndrome. However, she never fit neatly into any category. She was always one of a kind.

Two important events changed me forever. To this day, my spirit stays afire to keep the Rett movement alive—just so this will *never* happen to others. One is the young doctor who told me to just give up, saying that Stacie would never know what it was like to be normal and that I had to accept that and try to keep my own life normal. He said I must give up. The other was a child psychologist who told me that it was possible I had caused the “autism,” according to an outdated theory on autism that blamed “cold” mothers in the face of no other explanation for their child’s regression. You can be sure that these were the first two people to receive information about RS. Their insensitive comments launched me on a journey to prevent others from ever having to hear the same unfortunate advice. It was a broken road that led me to the diagnosis of RS, but the path forward from that news has been rewarding and hopeful.



Kathy and Scott Hunter. Below, the Hunter family



Since IRSA began more than twenty years ago on my kitchen table, I have longed for this book—a tangible resource to put into the hands of families new to the diagnosis of RS. Usually exhausted from the diagnostic process, they ask in so many different ways, “So, where do we go from here? What do we do while we wait for the cure?” As Stacie was growing up, there were no answers for our questions. It was important to direct energy at finding those answers and giving them to families everywhere as they arrived in “Rettland.” Authors are often told to keep the reader wanting more. *The Rett Syndrome Handbook* does not follow this rule. It was created for those who want it, and deserve *it all*. Hopefully, it will give you answers, spark your thoughts, fire up your imagination, and provide you with as much information as possible to give your loved one the best life she can have. You are where I was more than thirty years ago; to be able to give you this leaves me with remarkable rewards and immeasurable happiness.

Some of the advice in this book is sprinkled in more one than one flower bed. If you find the advice repeated but worded differently in more than one chapter, that’s because like fertilizer, it needs repeating. Just as RS has many stages, coming to terms with RS comes in stages. You are not done with the emotional stuff when you finish “Welcome to Rettland.” Keep turning the pages, and when you get stuck in a hard place, take pause ... then read on. You will find that the wisdom and insight of others who blazed the path before you cultivated some rich soil that will be like salve on your tender heart.

There may be forks in the road, a number of bumpy spots, and some rough obstacles in Rettland. But for all of the low places, there are magnificent high places to round out the view. With the experience and suggestions of others who have already been there, we are able to map out our journeys. We can be safe in the knowledge that we are not alone. It may take some time to adjust to the new scenery in Rettland, but once we fully arrive, the vista is spectacular.

Your experience with Rett syndrome will be a lasting journey of love. It may take awhile to realize, but you’ll come to know it in time. As you listen gently to the words that come from the hearts of those who have been there, you will recognize how much we share.

THE BOOK OF LIFE

It's too bad that the Book of Life doesn't come with a first draft so you can edit out the bad stuff and double the good stuff. But it doesn't work that way. You didn't choose to have a chapter on RS included in your life. Nevertheless, it is a part of you that you can't edit out or white-out. This is the plot you got. You have to make your own happy ending. Whether your Book of Life is a tragedy, a horror story, a drama, or a true life adventure is all up to you.

And you can do that because it's not what happens, but how you choose to *look at what happens* that makes your life what it is. Attitude is paramount. Every experience can be seen as an obstacle or an opportunity, depending on your outlook. You can choose to remain *bitter* or you can choose to get *better*. You can *complain* because rosebushes have thorns, or you can *rejoice* because thornbushes have roses. Life is more than what you are handed. It's what you *do* with what is placed in your hands.

Do you ever wonder why something can happen to two different people with radically different outcomes? I have two lifelong friends who had troubled upbringings. Both were children of alcoholics. Both grew up poor. Both became teachers. One committed suicide and the other built his own successful business. Each had the same difficult childhood, but one was not defeated by it.

You can't choose how you are born or how you die, but you can choose *how you live*. Your attitude is your most important possession. To say it another way, what happens *to you* is less significant than what happens *within you*. I often hear from parents when they are at their lowest point, just having learned their daughter's diagnosis. Many times I hear them say, "I don't know what else can happen." I say gently and quietly, "a lot." And the truth is, Rett syndrome is not the worst thing that could happen to them. Someday, down the road after it has had time to settle, they will realize that there are worse things than the diagnosis of RS. A lot worse.

WE WITHER

When you hear the words Rett syndrome for the first time, they bring on bewilderment, confusion, and sadness. Your heart droops, your hopes shrink, and your spirit wilts to the ground. You go through the universal stages of grief, no less than if your child had died. You may wonder what you did to make this happen and stay awake at night wondering how to make it go away. And then you have to face a couple of universal truths: 1) you didn't make it happen and 2) it won't go away.

Then, you get angry. Real, deep down gut-wrenching angry. Angry at God, angry at fate, angry at life. But you pull yourself together and keep moving forward, doing what needs to be done. Pretty soon you realize that there are not enough hours in the day to do what you need to do, and guilt enters the picture. There are so many competing emotions it's sometimes hard to separate them out. Many days, it's hard to see a patch of blue sky through the heavy clouds and there seems to be no silver lining, much less one you can see.

Over each door to the Rett family home could hang a sign with the Chinese proverb: "No one can say of this house, there is no trouble here." RS is hard. It hurts. It bewilders. It confounds. It defies reason. It shouldn't be this way. You would gladly trade a king's riches to take away RS. It will take some time before you realize that, just as Helen Keller said, "When one door of happiness closes, another opens; but often we look so long at the closed door that we do not see the one which has been opened for us."

Many times, I've thought to myself that I'd rather be a rotten person than have to deal with all this growth and greater wisdom stuff that comes from the suffering. But I can't change what RS has done to my

daughter. I can only change what it does to me. If I don't choose the growth and greater wisdom stuff, I'm guaranteed to wither.

The playwright Henry Miller wrote, "Life has no other discipline to impose, if we would but realize it, than to accept life unquestioningly. Everything we shut our eyes to, everything we run away from, everything we deny, denigrate, or despise, serves to defeat us in the end. What seems nasty, painful, evil, can become a source of beauty, joy, and strength, if faced with an open mind. Every moment is a golden one for him who has the vision to recognize it as such." Those golden moments come in time, when we learn to thank God, thank fate and be thankful for life. We learn that happiness does not require perfection and performance, only acceptance and love. This awareness does not come easily. We don't wake up a few days after the diagnosis and have a revelation. We meet hurt after hurt, challenge after challenge, and crisis after crisis. Then we are able to put a real name on hurt, on challenge, on crisis. We learn that the big things in life really are, after all, the little things.

Each of us has to learn to change what we can, accept what we can't, and go on. Those of us who have been crushed can attest to this: we grow stronger in the broken places. We gain so many valuable insights and gather so much courage. As one mother told me, "I am no longer afraid of anything." At the same time, we learn the true meaning of unconditional love and see the enormous power of the powerless.

Don't you find it amusing what other people think is a problem? It makes me chuckle out loud sometimes. I just smile and remember my own philosophy about problems ... "if little kids don't die from it, it ain't a problem." When I've been in line a long time and have exercised incredible patience with delays or bumbled transactions, the clerk often thanks me for my composure while others are flying off the handle around me. I usually reply, "Well, you know, I learned patience in the Hard School. I have a child with multiple handicaps. She can't do anything for herself. I've had a lot of practice. And you know what? This is small stuff. In fact, if this is the worst thing that happens to me today, I'll still be way ahead." It usually brings introspection to others who take a moment to reconsider what's important and what's not.

People have always amazed me when they comment about my patience with Stacie. I find it amusing to begin with, because Stacie is the one with patience. She has to wait to be fed until I think she is hungry, put to bed when I think she is tired. Too often, she has to put up with people who either act like she isn't there or treat her like she's an infant. People often remark that they "couldn't do it." I usually smile and say, "Well, what would you do?" Of course, love does not diminish with the diagnosis. It grows.

W E G R O W

We don't grow overnight. We grow in little spurts. We don't learn it all in books. Experience is the best teacher. When my first child was born, I looked at him and said to the doctor, "I've never been a mother before. I don't know what to do." The doctor smiled and said, "You'll be fine. He's never been a baby before, either."

The story I am about to tell is true. While I was pregnant with Stacie, I took a college course called Psychology of the Exceptional Child. For some reason, one phrase jumped out from the course textbook with the chilling words, "autism is the most severely debilitating disorder of childhood." I shuddered as I thought about what a wrenching experience it must be to have such a child. Then, with my vast experience as mother of two healthy youngsters and my newly found education from this informative course, I decided to write my final paper on "How to Counsel Parents of Disabled Children." I read all the right books, brought together the needed resources, and wrote the paper. I figured I must have said all the right things. I got an "A" on the paper. I was excused from the final exam because I was giving birth, but my good grades earned me an "A" in the class anyway. I thought

I knew everything there was to know. Then, about ten years later, I was cleaning out the basement when I came upon a box of old books and papers. There it was staring up at me: "How to Counsel Parents of Disabled Children." My mouth dropped open as I stared down at the "A" paper. I started to read what I had written, and then with sudden rage, I tore it into a thousand pieces and threw it in the trash. Bitter tears flooded my face as I realized how prophetic it was that I would choose this subject so long before, and how preposterous that I could even begin to pretend to know what to say before I had lived through it.

I think this is why as parents, our souls are so connected. Others can think about it, study it, teach about it, and write about it, but they don't live it. They don't really know it. We are a fraternity, a sorority, blood brothers and sisters, unrelated, but so very related. We come from many roads away and much distance apart, speaking any number of dialects, and observing different customs but we are one. An old Swedish proverb tells us that "shared joy is double joy, and shared sorrow is half sorrow." That's what joins our hearts. It hurts me to know that your child also has RS, but it helps me to know that we can get through it together.

We grow from this experience. Like the trees, we learn to bend with the wind before the force breaks us. And like some of the most beautiful flowers in the garden that bloom even more brilliantly after their blossoms are pinched off, we grow more from living with adversity than when everything is "perfect." The lucky ones learn to count their *blessings* instead of their burdens. Have you ever wondered what it would be like to have a child with RS and live in a country where there was little medical care and no special education? No family to help out? It's hard to imagine how some people survive without some lifesaving items we all take for granted, like disposable diapers, washing machines, Velcro, VCRs and DVD players, McDonald's French fries, Walkmen, IDEA and of course, Barney. We can't count our burdens without remembering our blessings.

B L O O M W H E R E Y O U A R E P L A N T E D

No matter what the circumstances, we can choose to be *alive* , to *thrive* , and to *survive* . Choose to grow and bloom where you are planted, as the saying goes. Throughout it all, it is important to always keep your sense of humor. Laughter gets the blood bubbling, swells the chest, jolts the nerves, sweeps the cobwebs from the brain, and cleans out the whole system. Laughter is physical therapy for the soul. You've come a long way when you can laugh at some of life's predicaments. It's good that we can laugh. And it's healthy for us to cry now and then.

R E A C H F O R T H E S T A R S

Above it all, it is important to hope, and I'd like to share my own hope with you. I hope to soon be out of work. I hope to make RS something no one ever heard of, not because it is *unknown* , but because it is *conquered* . I hope the next book I write will be titled, RS: The Cure. My hopes are very high. Once I didn't even have ordinary hope, and now I have "high, apple pie in the sky" hope. Hope is faith, reliance, and trust. We have every reason to be hopeful. We are so close to finding answers. It hasn't been easy. We've seen the misfortune of RS strike more than once in a family, but have seen the fortunate turnaround of families willing to overlook their own tragedy to participate vigorously in research. We have seen the sadness of girls lost to early death, yet have had our hearts warmed by parents who consented to autopsy so that the rest of us could be spared the same anguish. We've had bake sales, craft sales, car washes, races, dances, dinners, auctions, and concerts for the cause. It all adds up to hope. We are closer than we have ever been to solving the puzzle of RS. Just when you are about

to give up or lose hope, remember that the world is round—the end is often the beginning. We are going to come full circle. It's going to happen. We are joined hand in hand, by Carl Sandburg's promise, "where there is life, there is hope."

BELIEVE IN YOURSELF: BELIEVE IN HER

The insights we gain on the Rett journey are invaluable. While some may see our daughters as powerless, we know of the enormous power they have to change us. She may not walk, yet she helps us walk taller. As she struggles to move, she moves many hearts. She may need to be fed, yet she feeds our spirits. We seek to understand her, yet in so doing, we begin to understand far more. She may not speak to us in words, but she speaks to us in the silent language of love.

Your daughter brings so many blessings. She may never talk, but she will never talk back. She may never run, but she will never run away from home. She may need drugs to sustain life, but she will never take drugs to escape life. She may never use her hands for skills, but she will not use those hands for violence or evil. Look into her eyes and you can't help but have hope. She lives through her eyes. She loves through her eyes. As Dr. Alan Percy says, "The eyes have it." Her eyes, windows to the soul, take us to cherished places within her heart. Her eyes do more than see us. They *touch* us. Every human emotion is whispered, sung, shouted by her dancing, sparkling eyes.

While there may be confusion in her head, make no mistake, there is a lot of understanding in her head. She may have many obstacles to learning, but she can learn many things. Just because she can't *show* it doesn't mean she doesn't *know* it. It's in there. She's in there. Keep looking. You'll see.

It is most appropriate that this is called a "handbook," for hands are the trademark of RS. It is with our hands that we touch, feel, grasp, stroke, carry, comfort, and we give and receive. When we hold hands, we connect, and we double our strength. Our goal is to join our hands together, extending the circle ever wider until the day when others ask, "What *was* RS?" I could continue to write forever, for no book on the subject could include everything. For now, I hope this book lights your path.



Kathy Hunter
Parent, Founder and President, IRSA

To Julianna

My daughter is my mentor, my daughter is my strength.

My daughter is the center of my world, each and every day.

She taught me how to laugh, when times get really tough.

She taught me how to say "I'm sorry" when I say things that aren't okay.

She has taught me not to dwell on every little thing.

She has taught me how to be happy, when inside I felt like dying.

She has taught me there's more to life than perfect, normal or sane,

things like beautiful, stunning, surprising, and great.

She has taught me to hold my head up high,

to not be ashamed, to not want to cry.

She has taught me to love unconditionally, wholeheartedly, and sure,

to thank my lucky stars above, for a special daughter like her.

- PAM JORDAN

Special note to readers: For the sake of continuity and space, and as RS occurs almost exclusively in females, individuals with RS in this text are referred to in the female gender. It is acknowledged that RS can occur occasionally in males.

