Chapter 1
Visit 1: Part 1
"Jacob has autism."

The Diagnosis

After nearly two hours of evaluating Jacob (age 2 years 8 months), I was certain that he had autism (see Appendix 1 for the initial office evaluation). I had carefully questioned his mom and dad, Jim and Judy Grant (not their real names), about Jacob's language, social skills, and interests as well as the family's genetic history and Jacob's past medical history.

I had observed Jacob and his younger brother, 15-month-old Charlie, playing in my office and noticed that the brothers hardly interacted unless Charlie went after Jacob's trains. Then Jacob would become anxious and angry and aggressively push Charlie away. I couldn't recall Jacob seeking his parents to play even once through the hours of the visit. He went to them occasionally to pull them by the hand to leave but when he was told 'Not yet.' he returned to playing with the trains alone, head down, without referencing anyone. He put the various Thomas the Tank Engine trains on the wooden figure-8-track, pushed them around and around and flapped his hands in excitement.

Here is an edited version of the taped evaluation I made with Jacob's family as we neared the end of our visit.

Me*:* "Okay, let me summarize what I'm hearing you say. Jacob has a couple of dozen words that he uses inconsistently at the age of nearly three. This represents a *language delay*, but I like the fact that he understands when you announce routines like 'time to eat' or 'time to take a bath' and he is beginning to understand when you tell him to stop when he's doing something you don't want him to do."
Dad: "If he wants to."
Mom: "Otherwise he'll ignore you totally. We were worried about his hearing."
Me: "When it comes to his social interaction, he mostly goes into a corner in the daycare and plays with toys alone and doesn't play with his peers. I also notice that he doesn't interact much with his brother. And he hardly referenced you to play with him today, which you say is pretty typical for him. So he has problems with *social engagement and interaction*.

Also, he loves cars, trains and trucks and watches how the wheels move. He lines his trains up. He opens and closes doors a lot. And he's a big fan of Baby Einstein videos. So he has *dominating and repetitive interests*."
Mom: "And he'd watch those videos all day long if you let him."
Dad: "Which we don't."
Me: "Finally, he has several *sensory issues*. He puts his hands over his ears when he gets upset and when he gets upset he can stay upset for a long time. Loud, chaotic, noisy environments bother him. He does a little toe walking and flaps his hands in excitement. He is very visual and loves to look at lined up objects. And he is not a great eater because he likes food that's smooth.

I'm so sorry to say this but I'm afraid Jacob has autism. It's a *mild* form of autism but I feel quite certain that he has it. "

I stop talking to give the diagnosis time to sink in. Judy Grant's face clouds up and tears form. I offer a tissue. Jim Grant's hand reaches over to hold his wife's hand; his eyes go down; and his face losses all expression.

Me: "I'm sorry."
Mom: "I knew it."

Dad: "We've been waiting to hear this for a long time now."
Mom: "It's just hard to hear."
Me: "No parent wants to hear that anything is wrong with their precious child. . ."

The Path to Diagnosis
Jacob's parents came to my developmental and behavioral pediatric practice after a series of frustrating events.

When they first suspected that something was wrong with Jacob's development at 18 months, they rushed to the Internet where they found an overwhelming amount of information that led them to believe that children could be cured of their autism through the use of special diets, vitamin shots and other 'biomedical' therapies (Chapter 4).

Then they went to their primary care doctor who, sadly, told them that boys tend to develop more slowly than girls and to 'wait and see' how his development goes over the next 6 months. (See *Resources*: AAP recommendations at the end of this chapter). And the grandparents on Jim's side of the family confirmed that even Jim developed language later than his sister, so not to worry.

By the time Jacob was two and still not talking, the Grants became desperate to find help. Eventually (and sadly from my perspective) after almost a year of worrying about Jacob's lack of language they heard about my practice from an acquaintance. Their story is not unusual at all.

Accurate Information
In this first visit with Jacob and his family I feel the need to cover the first essentials: *what autism is*, how I came to my diagnosis, and where the parents can find *accurate information about autism*. I make sure I give the Grants time to talk about their reaction to the diagnosis (see Chapter 2) and then we discuss intervention options.

The Grants are recovering from the initial shock of my diagnosis. The tears are drying and Jim and Judy Grant just look drained. I take out my old style cassette tape recorder to record our discussion because I know that for the rest of the visit the Grants wouldn't be listening to much of what I had to say but their own grief.

Me: "Do you mind if I record our discussion? This way you'll have a document of important information."
Dad: "That's fine, that's fine. . .So what do we *do*?"
Me: "The bad news is that Jacob has autism; the good news is that with proper intervention I think he could do very well."
Mom: "I'm so upset."
Me: "Listen, mom. Jacob has some language before age three. He has no unusual physical findings. He seems smart. I'm very optimistic. Let me start by giving you a little overview of what autism is and what we have to do next to get you on the upward path."
Dad: "Sounds good."
Me: "The first thing I want you to know is that *this is not your fault*."
Mom (tearing up again): "That's a big relief. I was worried I had done something wrong."
Me: "No. This is not due to parenting or what you ate or did during pregnancy. And, mom, you told me everything went well during Jacob's birth. So there is nothing medical that caused Jacob's problems."
Dad: "So what did cause this?"

Autism is A Genetic Condition
Autism is a genetic condition that causes poor nerve connections in the brain.

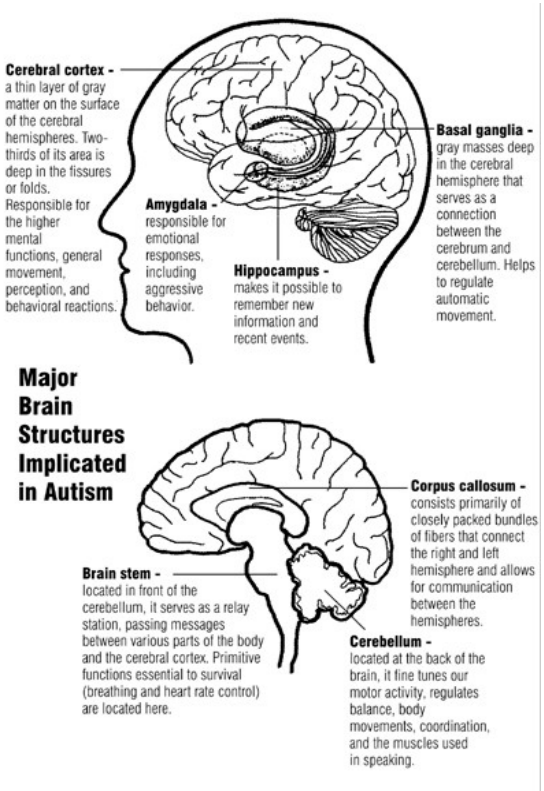
There are 20+ autism genes and solid scientific evidence has found that the relatives of children with autism have some of the traits of autism like mild language delays or being socially shy or being very precise and analytic. When I take family histories and look at the 'family tree' I very frequently find certain jobs over-represented including engineers, computer technicians, mechanics, tool-and-die workers, accountants and other 'left-brained', linear thinkers. Other common traits found in relatives of children with autism include: obsessive-compulsive tendencies, language delays, and relatives who are 'collectors', who were 'odd' socially and those who never succeeded in the workplace.

Importantly, if a family has one child with autism there is a 5-8% chance that a second child will have it. In identical twins the chances are 90%! That autism is a genetic disorder is not in question. Over 500 articles on the genetics of autism were published this year alone. (See *References & Resources*: *AGRE*).

Me: "In fact, dad, when I was asking you about your family tree you mentioned that your uncle, who was a tool-and-die worker, was not very social at family gatherings."
Dad: "Odd as a three dollar bill."
Mom: "I'm very perfectionistic myself and Jim you have to be very precise in your work. (Dad was a high-level information technology (IT) consultant.)
Me: "I like to say that when smart people marry smart people the odds of having a child with autism goes up."

The Underconnected Brain
Autism genes cause the brain of the child with autism to be 'under-connected'. (See *Resources: Autism and The Brain*) What I tell parents is that *their child's brain is like a net that is too loose and can't capture the complexity of the world.* Typical children with typical brains seek novelty; children with autism seek 'to keep the world the same' which sadly keeps them in their own world. This is why they do the same things over and over. It makes a chaotic world predictable. It makes them feel comfortable, less anxious, less overwhelmed. I call this the child's *'neurologic comfort zone'*.

Dad: "Then autism is a brain condition that makes it hard for our kids to connect so they stay disconnected."
Me: "Well said, dad. Kids on the spectrum have a hard time with anything that's too complex. What are our most complex functions? Language and social skills. And these are the main deficits in autism. The job of intervention is to tighten that 'neurological net' so the child can make more sense of the world, gain language and be social."



Scientific Note (Notes in small print can be skipped): 'Autism genes' code for proteins in the

brain that typically help make strong nerve connections. In autism, these connections are weak. Brain areas (**see Figure above**) affected include the *frontal lobe* where decision making happens; the *cingulate gyrus* where impulses are kept under control; the *hippocampus*, which makes it possible to remember new information and recent events; and the *basal ganglia* which helps to coordinate automatic movements. In the *amygdala*, which is the emotional center of the brain, there are too many neurons but they are stunted and do not branch appropriately. This is why children with ASD have problems with understanding other people's emotions. The *cerebellum*, which controls fine and gross motor coordination, has fewer neurons in autistic brains, which may explain the common signs of hand flapping, toe walking, bizarre movements, etc. Finally, the *corpus callosum*, a large white matter mass that coordinates activities between both sides of the brain, tends to be thinner in children with autism. This points to poor coordination between as well as within areas of the brain.

Interestingly, because of these brain abnormalities, the brain of the child with autism is actually *enlarged* early in life resulting in an *increase in their head circumference, which* has recently been accepted as a neurological sign of autism. In most children in the head circumference normalizes by a later in childhood.

When we look less at the anatomy and more at the physiology of autism, research is now showing abnormalities on functional MRI (fMRI) scans which look at areas of oxygen consumption in the brain tissues. Here the findings are consistent. People with autism have trouble—there is less metabolic activity in the area of the brain having to do with—understanding facial expressions and reading other people's emotions. They tend to pay attention to unimportant social cues. They prefer looking at objects instead of at people's eyes, for example. Children with autism are very visual, which is why they often stimulate themselves visually (e.g. lay their heads on the floor and push train in front of their eyes) and think in pictures.

Dr. Nancy Minshew, a nationally regarded child neurologist, and her team, using neuropsychological evaluations, has consistently found that people with autism have difficulties on all sorts of complex tasks not necessarily related just to language and social skills. They may have problems with handwriting or speed of doing a task. They tend to be 'concrete' thinkers and take things literally. So when you tell them to 'hop to it', they literally hop. Abnormalities in the frontal and temporal lobe lead to difficulty with what's known as *theory of mind*, which is the inability to take other people's perspectives. In other words, they don't have a *theory* that other people have a *mind* of their own.

Why is Autism Increasing So Much?

Me: "In short, on the basis of brain science, children with autism have difficulties with the two most complex of human abilities, namely, language and social skills. They want to keep the world the same, which leads to repetitive and dominating behaviors and interests. These three characteristics—delays in language, delays in social skills and dominating interests—define the diagnosis of autism!" (See *Resources*: DSM V Diagnosis of Autism).

Dad: "Well, I was reading in the newspaper just yesterday that autism is becoming an epidemic. What I don't understand is if autism is genetic as you say then how come it's keeps increasing so much. I never heard about an epidemic of Down Syndrome."

Me: "Very astute, dad. The people who study population trends in autism have concluded that this increase cannot be due just to genetics alone. While most experts agree that a big part of the increase in autism is due to the practice of diagnosing children earlier and diagnosing milder cases, something else is going on. In fact, we're now at 1 in 68 children. And with boys four times more likely than girls to get autism, we're at 1 in every 42 boys!"

Mom: "Why is that?"

Me: "Males are more linear in their thinking. It may have to do with testosterone. No one knows for sure. I just say that 'women are the superior species'." (All laugh.)

Dad: "But why the big increases. I'm seeing road signs that say *By the time you get to work, a child will be diagnosed with autism*."

Environmental toxins and Autism

Me: "I don't want to take up too much of our time now but the continuing increase in autism now appears to be due in part *exposure to toxins while the baby is in the womb."*

Mom: "Toxins in the womb?!"

Me: "In an effort to explain the dramatic rise in autism, very recent research (See References) has found that *babies' brains while they are in the womb are exposed to hundreds of neurotoxins* like dioxins, PCPs, fire-retardants, and especially pesticide residues, among others. When these babies also have the autism genes then together neurotoxin exposure plus genetic tendency combine to cause the under-connected brain of autism."

Mom: "Could I have avoided this by eating better?"

Me: "No, it's in the air, the food, the water. There's really no avoiding it. We're all contaminated."

Dad: "That's really sad."

Me: "It appears that most children have to have *both* the genetic tendency and the toxin exposure in order to get an ASD. What has also become clear is what does NOT cause autism. It is *not* caused by immunizations (See *Resources: Autism & Immunizations: The Facts*); it is *not* caused by mercury poisoning. It is *not* caused by diet or allergies.

Other key factors may be more physical than genetic (See *Resources: Autism and Environmental Factors*)

Factors That Increase The Risk of Autism

- In utero (in the womb) exposure to toxins
- Being an older mother
- Being an older father
- Changes in father sperm cells
- Maternal obesity
- Taking SSRI's (Prozac-like) medications during pregnancy
- Extreme prematurity
- In vitro fertilization
- Others

In vitro fertilization (IVF), having an older father, older mother, and having a complicated, premature birth are all associated with an increase in autism."

Brain plasticity: There is real hope

Despite the problem with genes, toxins and environmental factors, children with ASD luckily have something called *neuronal plasticity*. Children's brains are very 'plastic', growing at an astounding rate in the first seven years of life. Brain size doubles by age three and ninety percent of brain size is reached by age 7! In essence, brain plasticity is the ability of the brain's nerve cells to make many new connections to be re-shaped from the outside in, through effort and experience. This is especially true of autism compared to other developmental disabilities. This is what makes children with autism different and this is why children with autism need intensive intervention as early as possible.

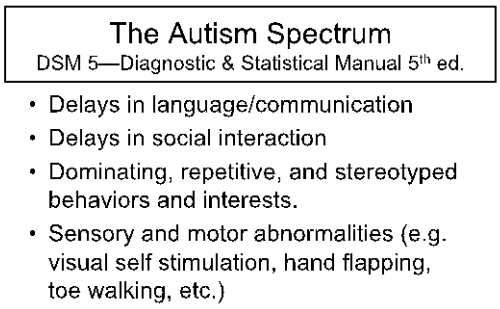
Me: "So, while Jacob's autism is not your fault, still there is a lot we can do to help him. Before we get into intervention though let me talk about where Jacob is on the spectrum."

Autism Definition

As I explain to the Grants, autism spectrum disorders (ASD) are defined by delays in language/communication, delays in social interaction and having dominating interests and behaviors. Sensory and motor abnormalities are also common. This is the definition used by psychiatrists and psychologists and can be found in the Diagnostic and Statistical Manual 5th Edition also known as the DSM 5.

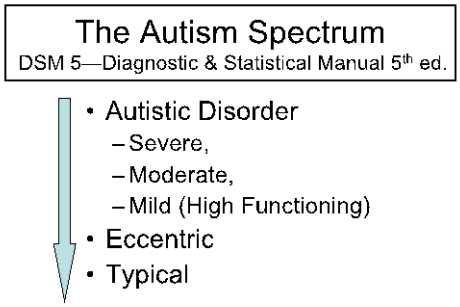
Jacob's parents were thrown off the diagnosis by paying attention to exceptional behaviors. Jacob's dad said: "But Jacob makes eye contact and is very loving."—He didn't see the big picture of

delays in language, poor social interaction and dominating and repetitive interests.



Also confusing to parents is the broad range of severity of the autistic spectrum disorders. Children can have no words, very poor social interaction and very dominating behaviors (e.g. lining up toys, opening and closing door, etc.). These children would be on the 'severe' end of the spectrum.

Children can have *almost* normal language, some difficulties with interaction and dominating interests (e.g. cars, trains, superheroes). These children would be of middle or moderate severity on the autism spectrum.



Then there are those who have fairly normal language but are still socially inept or very awkward and have dominating intellectual interests (e.g. dinosaurs, planets, Star Wars, etc). These are children would have a mild autism spectrum disorder and be described as having high functioning autism. In order to be on the spectrum, however, there must be *significant social impairment*. As children improve they may appear eccentric but quite functional socially (a common term still is use for these children is *Asperger's Syndrome*) and about 10% of children with autism can actually outgrow their condition! (See *References*)

These variations can make it hard to diagnose a child's condition. That's why it's very important to get good professional evaluations. (See *Appendix 2*: *Screening & Diagnostic Tools*) Recent research concludes that children can be diagnosed with an ASD as early as 14 months (See *References*).

In some states the school system can make an 'educational' *diagnosis'* but usually it's best to go to those with experience like a developmental/ behavioral pediatrician, neurologist, pediatric psychologist or child psychiatrist.

Me: "I'm actually optimistic about Jacob. He has some words, he can be fairly easily engaged and his self-isolating behaviors are not too severe. *So, he has a mild to moderate form of autistic disorder with a good prognosis."*
Dad: "So you're saying Jacob has real autism not high functioning autism."
Me: "Yes, for now but with intensive intervention children on the spectrum can really make amazing progress. I see it all the time and Jacob could be one of those children. I don't have a crystal ball but I'm very hopeful that he will make very good progress.
Mom: I read recently that children can be cured of autism.
Me: Well, I never say 'cured' but there was a study that found about 10% of children 'outgrow' their symptoms and did not meet the DSM 5 criteria. I have many cases of children who just kept getting better to the point where they

functioned typically in school and made real friends."
Dad: "I didn't know that was possible."
Mom: "I'm afraid to ask but could Jacob be in that 10%?"
Me: "It's possible but we won't know until we get started with intervention which I want to talk about next."
Dad (looking sadly at his wife): "We'll do whatever it takes."
Me: "I know you will. But let me turn this tape off and let's talk a little about how you guys are doing."

Visit Summary:

- The members of the Grant family—Jim, Judy, two and half year old Jacob, and fifteen month old Charlie—are introduced.
- I review Jacob's profile and diagnose him with *mild to moderate autistic disorder* (See also Appendix 2 for my report on Jacob).
- The causes of autism—genetics, toxin exposure in the womb, and other environmental factors—are described.
- I define 'autistic spectrum disorder' according to the official criteria and explain why it is increasing so much.

References & Resources:
For the best information on what autism is I recommend the following web sites and books:

Websites:

- *Autism and the Brain*
- *Autism and Environmental Factors*
- *AAP Autism Toolkit:*
- *Autism Speaks 1st 100 Days Kit*
- *University of Michigan Your Child* website
- *First Signs.org*
- *Autism Genetics Resource Exchange (AGRE)*
- *DSM V Diagnosis of Autism*
- *Autism and Immunizations: The Facts*

Books

- *Could it Be Autism? A Parent's Guide to the First Signs and Next Steps*, by founder and president, Nancy D. Wiseman (Broadway Books, 2006).
- *The Autism Book* by Jhoanna Robledo and Dawn Ham-Kucharski. (Avery/Penguin Books, 2005)

Coming Up Next:
In Chapter 2, I explore the common emotional responses families have when I make the diagnosis. Then, in Chapter 3, I'll talk about what I advise parents to do when they leave my office and begin intervention; that is, what I would do for my child if he or she had an autistic spectrum disorder.