

CHARGE Association: A Personal Account
Tim Hartshorne
Central Michigan University

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Not every school psychologist has the opportunity to see life from the other side. It is not something that I would necessarily have chosen to do, but it has been an incredible experience. Our son Jacob was born May 10, 1989 with what is known as CHARGE Association (or sometimes Syndrome).

The Beginning

Prior to a few weeks before he was born, the pregnancy had gone smoothly, and there were no indications of problems. Then, eight to nine weeks before the due date, Nancy had contractions. After two weeks of bed rest and medication, the baby decided to come. It was a busy delivery. Our family physician attended, along with her specialist, the neonatologist, two residents, two nurses, and myself. There was no placing the baby on its mother's stomach, or anything else. The baby was out, and immediately he was placed on an intensive care cart while the doctors worked on him. I had no idea how he was doing, and just tried to interact with Nancy. What they were doing was intubating him, or placing a tube down his throat, and stabilizing his breathing. Then he was whisked away to the Neonatal Intensive Care Unit (NICU).

The NICU

Jacob was one of about four babies in NICU. One was very tiny, another was very sick (and died during Jacob's stay). It was a bit of a shock to see your child with wires coming out of every which way. Jacob was 4 lbs. 10 oz., and looked very tiny lying on his bed. One of his nurses the first night made a little card with a turtle on it that said, "Welcome to Jacob Holmes Hartshorne." That was really nice.

In fact, the nurses were really nice. They seemed to understand what we were going through as parents, and tried very hard to talk to us, answer questions, let us know how he was doing, and as much as possible let us hold him (which we were able to do after about three days). The nurses encouraged us to participate in his care, and I changed a number of diapers, and learned to give percussion (a form of respiratory therapy), but left the bathing to Nancy.

There were two neonatologists on the NICU. They were both excellent, although they had differences in style, and also apparently in the way they viewed the infants. Ours was the more conservative, and for example felt that Jacob was at least 10 weeks early, while the other thought Jacob to be only six. They compromised for the record on eight.

Other than prematurity, we had no indication at first that there were any problems with Jacob. The various doctors assured us that premature babies tended to do well, and Jacob should be fine. The initial problem he had was difficulty breathing, which is common in premies. Jacob's problem was that he did very well with the breathing tube, but then when they extubated it (took it out), he began having problems breathing.

While his breathing problems remained a mystery, the neonatologist informed us that, on closer examination, he had noted a number of anomalies. For example, Jacob's ears were low, rotated, and misshapen; he had some extra skin between his eyes; his penis was small; he had a simian crease on his palm. He told us that while one anomaly like that is often not a problem, to have several might mean something else was wrong.

This was not easy information for us to absorb. But it was only the start. We went through three chromosome studies, trying to get one that would grow. The third indicated the chromosomes were normal. We celebrated. However, in the meantime we learned that Jacob had a hole in his heart, although the heart specialist reassured us many times that it was small, and not something to be alarmed about.

The most difficult information came while I was out of town for a job interview, and while our neonatologist was attending a conference in Alaska. The other doctor attempted to place a feeding tube down Jacob's nose. The tube would not go through either side. The passages in the back of Jacob's nose were blocked.

The Diagnosis

While we wanted Jacob to come home, we were also nervous about it. But after about five weeks the hospital decided we were ready to take over. His breathing was stabilized by the use of a bottle nipple with a hole cut in the end. The nipple was then taped into Jacob's mouth. Since babies are nose breathers, this device kept Jacob's mouth open all the time so he would continue to breathe.

Before we took him home we were trained in infant CPR, and the use of a suction machine for clearing out Jacob's throat and nose which tended to become very full of mucus, and an apnea monitor to alert us in case he should stop breathing. We soon fell into a routine of tube feedings, suctioning, and percussion, along with the normal challenges of parenting a newborn.

We had only had Jacob home a few weeks when our neonatologist called. He reported that he had been doing some reading and come across something called CHARGE association that seemed to fit Jacob. It would be several months before we were able to have the diagnosis confirmed, but we knew immediately that it was correct.

CHARGE is an acronym. Children with the diagnosis vary greatly in their manifestation of each of the associated problems.

The **C** is for Coloboma of the eye. A coloboma is a missing piece. Jacob has retinal coloboma. The missing piece in his right eye is in the middle giving him very limited vision. In his left eye it is to one side giving him a blind spot, but more vision.

H is for Heart Defect. Jacob has a very slight hole that can only be seen by enhanced techniques. Because of its size, it should not cause him problems.

A is for Atresia of the choanae. In English, this means that the openings in the back of the nose that allow air to pass are blocked. Jacob's condition was severe in that both airways were blocked, and blocked by bone along with cartilage. This condition has been corrected through a series of surgeries at KU Med Center. The openings had to be drilled, and then plastic tubes inserted to prevent them from closing. They had to be enlarged every other month as he grew, and larger tubes inserted. His last tubes were removed when he was eight months.

R is for Retarded growth and/or development. CHARGE children may be retarded and/or short. Jacob is now 22 months. He weighs less than 20 pounds. Part of the problem may be due to feeding difficulties. He can only drink thickened liquids and eat strained baby foods. He vomits frequently, sometimes several times a day, at other times only once or twice a week. In terms of mental retardation, he does function at a low level, but his development is complicated by his vision and hearing problems.

G is for Genital hypoplasia. In boys, this means the penis is small and the testes cannot always be felt. This can usually be corrected through hormone injections when the children are older.

E is for Ear malformations. Not only are Jacob's ears somewhat low and rotated, but he is also profoundly deaf.

Services

Jacob receives services through the schools. His teacher comes to the house twice a week, and he receives PT and OT at the center once a week. He receives audiological services through the University clinic, and an audiology intern comes to the house once a week to work with a vibro-tactile aide. An infant mental health specialist is also consulting with us.

Perhaps our favorite service consists of volunteer students from the University. I teach an undergraduate psychological testing course that is primarily made up of special education students. I simply ask if any of them would be interested in working with Jacob, and generally get four to six volunteers. They love playing with him, and he seems to thrive on the added attention. It was a student who taught him for the first time to reach for his pacifier and put it in his mouth – a big step for Jacob.

We receive respite care in addition to a subsidy check from the State Mental Health Department. Michigan Crippled Children also covers all of Jacob's uninsured medical expenses. These things really help!

Jacob

Naturally, the clinical picture does not describe our son. He is cute, cheerful, and easy to be around. He has his preferred toys, frequently enjoys interaction with adults. And seems generally content. His fingers are very busy. He holds them in front of his eyes (a typical behavior of infants with visual problems) and makes signs. He is beginning to recognize that they are signs. While he cannot crawl or walk yet due to muscle tone problems, he does scoot around on his back. He helps with his dressing, likes to put a comb through his hair while his mother is combing it, and enjoys examining new objects.

All children are challenges for parents. Jacob presents some unusual challenges, but we do not view them as terrible. CHARGE is a very rare disorder. Its etiology is unknown, although it occurs during the first 30 to 45 days of gestation, and the children vary greatly in how severe any one of the characteristics is. Our orientation has been from the beginning to accept Jacob as he is, focus on what needs to be done on a day-to-day basis, and work to help him meet whatever his potential might be. There is much joy in that.