Mystery finally solved

Test reveals answer after genetics evolves with the years

By Mary Jo Hebert, Commentary
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Saturday marked National DNA Day, the day that commemorates the completion of the Human Genome Project in April 2003. The research project's goal was to map and understand all the genes of human beings. In the time between then and now, I have come to think of the mapping of genes as having all the pages of an instruction manual needed to make the human body.

Some manuals, it turns out, have missing or extra pages. Or paragraphs. Or sentences. Or words. Or letters. Still others contain the right content, but the order is out of sequence. When this happens problems often arise. Children born with extra, missing or re-arranged genetic material are born with rare genetic disorders.

When our son was born 23 years ago, much of the manual's content was still a mystery. Elated to welcome a third son, Louis, a beautiful baby with a soft cry, our excitement quickly turned to alarm when shortly after birth he suffered a seizure, and a CAT scan revealed brain damage from a cerebral vascular accident.

Floppy to hold, Louis' low muscle tone, called hypotonia, made feeding difficult and weight gain nearly impossible. Stamped on the cover of his medical chart were the words "Failure to Thrive." Didn't that refer to babies who weren't hugged enough? I asked, confused and desperate for a diagnosis that would explain his difficulties.

Every day brought more findings: A hole in the wall that separates the heart's top two chambers, a faulty valve that regulates blood flow, an abnormal opening of the urethra. But why?

Not everyone appreciated our need for an explanation, however. A cardiologist with little patience for questions asked what difference it would make; it wouldn't change a thing.

Birth defects, our pediatrician explained, are often suggestive of an overall genetic syndrome,

one that would provide a single explanation and connect the pieces of the puzzle. During our first genetic evaluation, after asking a battery of questions — uh, no, it is not possible that my husband and I could be related — a geneticist ordered a blood test called high resolution banding chromosome analysis.

When the result of the test came back normal, our hopes for a diagnosis were dashed. While chromosomes could appear normal, it was explained, what was happening on a more microscopic level, on the genes themselves carried by the chromosomes, was still unknown. Technology could not yet access the fine print on the manual's pages.

But then came the promise that renewed our hope: one day it would. In the meantime we would have to wait. And so we did. Twenty years.

In 2011, we learned about a test called SNP Microarray Analysis that detected genetic changes associated with a variety of syndromes. After years of disappointing dead ends, we braced ourselves for another letdown. One month later a genetic counselor called to say she had news.

Koolen de Vries Syndrome is a rare genetic condition in which a tiny piece — five genes and a part of a sixth — is missing from chromosome 17. Characterized by intellectual disability, epilepsy, and heart and renal defects, the syndrome provided a dead-on description of the difficulties our son faced. A page-by-page account of how and why things had gone wrong.

And to the cardiologist who asked what difference having an explanation would make, I'm here to tell him: all the difference in the world.

Let's start with this: It wasn't my fault. For years, I had agonized over the glass of wine I drank before I knew I was pregnant, the Tylenol I swallowed the day I couldn't stand the headache.

A doctor's letter I had kept from years before stated: "Mother denies gestational exposure to drugs, alcohol, and tobacco." To a mother already holding herself suspect, those words translated as, "Well, that's what she says anyway, but who knows what she was really up to." And then there was the issue of heredity. With two older children who may plan to have families of their own one day, we always wondered — could this happen again? Thanks to a diagnosis, we now know the likelihood of recurrence is extremely low. Similarly reassuring is the knowledge that a diagnosis can often be used to predict potential future problems before they emerge.

The revolution in technology that led to the completion of the Human Genome Project provided an avalanche of answers to scores of previously unanswerable questions. Answers that would provide me not only with an accurate manual, but an improved lens through which to view it.

The problem had never been with our son, after all. The problem had been with the manual. We had been using the wrong one.

For 20 years our son had been measured against standards dictated by a manual that did not apply to him. And always he came up short. Delayed. Below average. Less than.

Given the right manual, a rare and limited edition, I can now finally see him for who he is — not a flawed creation, but a masterpiece of design.

And that not only changes one thing. That changes everything.

Mary Jo Hebert lives in Clifton Park. Her e-mail address is hebertmaryjo@gmail.com.

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