Physician sees disorder for decade before genotyping can confirm genetic link

It was more than ten years ago that Dr. Tally Lerman-Sagie first saw babies with PCCA, a genetic disorder that causes severe mental and physical disabilities and brain atrophy—all before age three. Parents would bring in months-old infants with these unexplained seizures. Lerman-Sagie tested the babies as fully as she could in the pediatric neurology clinic she heads, just outside of Tel Aviv. At first, the test results would come up normal; only as the afflicted babies grew older would their MRI scans show their brain atrophy. Then, one family had a second child afflicted with the same mysterious symptoms. That made Lerman-Sagie realize she must be dealing with a genetic disease… but knowing that wasn't much help, either. She didn't know the gene, so she couldn't test potential parents for it. She certainly couldn't cure the disease.

Now, however, improving DNA technology has helped Lerman-Sagie discover the culprit genes behind PCCA, offering families a chance at prevention.

Read the full, original story: Hunting For The Genetic Secret To A Rare Disease