

# Mother passes on detectable genetic disorder to child

Neurologist had ruled out disease

\$10 million arbitration award

The case resulted from a child born with a genetic disease she inherited from her mother, who was unaware that she had the disease despite having presented to the defendant neurologist with signs and symptoms for 13 years. The plaintiff also was unaware of the 50-50 chance of passing the disease on to her child.

Beginning in 1992, when the plaintiff was 18, she began to experience minor seizure-like episodes. She saw several neurologists for her condition, but none came to a diagnosis.

In 1995, she was referred to the defendant neurologist and epileptologist. In 1998, early on in her treatment, the defendant ordered a brain MRI, which was suggestive of tuberous sclerosis, or TSC, a genetic disease causing non-malignant tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs and skin.

Eighty to 90 percent of those with TSC suffer epilepsy. TSC affects the central nervous system and can cause seizures, intellectual disability, developmental delays, autism, skin abnormalities and renal cell carcinoma. A person with TSC has a 50 percent chance of transmitting the disease to his or her child, who can be more severely affected than the parent.

The clinical criteria for diagnosing TSC, codified by an international consensus panel in 1998, recognize several "major features." The presence of any two of the major features warrants a "definite" diagnosis. Although the plaintiff fit the criteria, the defendant testified that he ruled out TSC in October 1998 based on "the absence of other clinical evidence to support that diagnosis."

The defendant continued to treat the plaintiff for 10 years with anti-seizure medication. He never determined the etiology of her seizures and noted he remained "uncertain" of what he was treating. He never performed an-

other brain MRI or other diagnostic test or referred her to another neurologist (including an expert in TSC who worked in the same hospital). He did not obtain the plaintiff's dermatological records, which included frequent visits to dermatologists who repeatedly noted red bumps on her nose and cheeks (facial angiofibromas), a major skin feature of TSC.

In 2005, the plaintiff had a daughter, who was born healthy. In 2008, while still under the defendant's care, the plaintiff gave birth to her second daughter, who began to suffer seizures and spasms at 5 months old. Testing confirmed that she had TSC.

At that time, the plaintiff's 1998 MRI was reviewed by other neurologists, who observed the cortical tubers and facial angiofibromas. They concluded that she, too, had TSC and that she had genetically transmitted it to her daughter. A renal scan then revealed that the plaintiff had a cancerous tumor on her kidney due to TSC, which subsequently was removed.

During the child's first several years of life, she suffered severe infantile spasms and seizures, which were difficult to control despite medication. The child is now 6 and suffers multiple consequences of tuberous sclerosis including autism spectrum disorder, ADHD, and cognitive, speech and fine motor delays.

The defendant testified that excluding TSC from his diagnosis in 1998 was appropriate and in conformance with the standard of care, as he had conducted a wide variety of tests including repeated brain MRIs and EEGs, as well as skin examinations, none of which was conclusive for TSC. He also maintained that the disease became truly diagnosable only after her second baby was born with it.

The arbitrator found that the defendant was negligent in failing to diagnose the plaintiff with TSC or take adequate steps to insure an accurate diagnosis by referring the plaintiff to a TSC expert. He found that the defendant



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negligently ruled out TSC given an MRI suggestive of TSC and the presence of facial angiofibromas, which are major features of TSC.

The arbitrator concluded that had the plaintiff been diagnosed with TSC in a timely manner and received appropriate genetic counseling, she would not have had children through natural conception.

Expert testimony from a life care planner and economist estimated the present value of the future medical, educational and other extraordinary costs associated with the care of the plaintiff's child to be between \$11.2 million and \$13.1 million.

**Action:** Medical malpractice

**Injuries alleged:** Wrongful birth; cognitive and behavioral developmental delays

**Case name:** Withheld

**Court/case no.:** Withheld

**Jury and/or judge:** N/A (arbitrated)

**Amount:** \$10 million

**Date:** Oct. 16, 2014

**Most helpful experts:** Dana C. Hewins, economist; Sandra Lowery, life care planning

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