Before the mid-1980’s, little was known about mitochondrial disease. Patients were likely to be misdiagnosed as having cerebral palsy, Parkinson’s or other disorders.

Such was the case with Emily R., a 13½-year-old girl from Ellicott City, Md., whose parents were told she had cerebral palsy. Emily, who exhibited symptoms at birth, is fully dependent and unable to walk, eat, or talk.

“Her disorder is in the muscular dystrophy family,” says her mother, Kathy.

Kathy is a former pediatrician who stays home to care for Emily and two other children who also have the illness but with milder symptoms.

Kathy herself has mitochondrial disease and says her symptoms include muscle twitches, GI problems, a weakened immune system and allergies.

John, 6, has autism and Crohn’s disease. Kelly, 16, has severe ADHD. Both suffer from muscular problems due to low muscle tone and weakness.

Through advanced research, scientists have learned more about abnormalities in the molecular powerhouse known as the mitochondria, which converts glucose and oxygen into energy.

But due to an inherited condition, the mitochondria can fail, causing cells to lose energy and become damaged and even die.

Last summer Emily was able to take two Angel Flights to the Children’s Hospital in Atlanta—first to be evaluated and diagnosed, and then to undergo surgery.

“She seemed to enjoy the flight,” Kathy notes. “The pilots couldn’t have been more wonderful.

There was no other way we could have gotten down there.”

Emily suffers from severe scoliosis and, following diagnostic tests, was deemed to be a good candidate for the spinal fusion surgery.

The complex seven-hour surgery required inserting rods and screws in her back.

“She was in the hospital for a week,” her mom says. “She is still on bed rest. It’s a long recovery.”

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