Genetic testing reveals previously unknown diseases

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis.

Simon Kunesh arrives at his physical therapy appointment hanging from his mother's neck, arms locked, swinging back and forth like a pendulum.

Simon is 14 years old. He has sandy hair, a sprinkling of freckles, a goofy smile, and ears that look like stick-on accessories for a Mr. Potato Head toy. His head is small for his age — the result of brain damage sustained from frequent violent seizures — but it fits his bony frame. Although he's steadily grown taller year by year (defying the scoliosis that's slowly twisting his spine into a lopsided S), he hasn't gained a pound since 2007. He stands four feet and weighs 40 pounds.

With a cognitive capacity roughly equivalent to a toddler's, Simon displays symptoms of a number of fairly common ailments, including cerebral palsy and autism. After more than a decade of tests, however, his doctors have concluded that his condition appears to be something new — something that hasn't yet been recorded anywhere else in the world.

Simon's situation isn't unique. Thanks to rapid improvements in genetic testing, scientists are identifying more and more diseases every day.

Though these ailments always have existed, because each one affects fewer than 200,000 Americans, the chances that a child like Simon would get a diagnosis 10 years ago hovered at about 3 percent, says David Craig, deputy director of bioinformatics at the Phoenix-based Translational Genomic Research Institute (TGen). While parents of such children shuttled them from specialist to specialist searching for answers, the vast majority were either shoved into a diagnosis that didn't quite fit or remained medical mysteries.

Read full, original post: Arizona mom discovers her son may be only one in the world with his disease