Research team helps father discover source of son's disability

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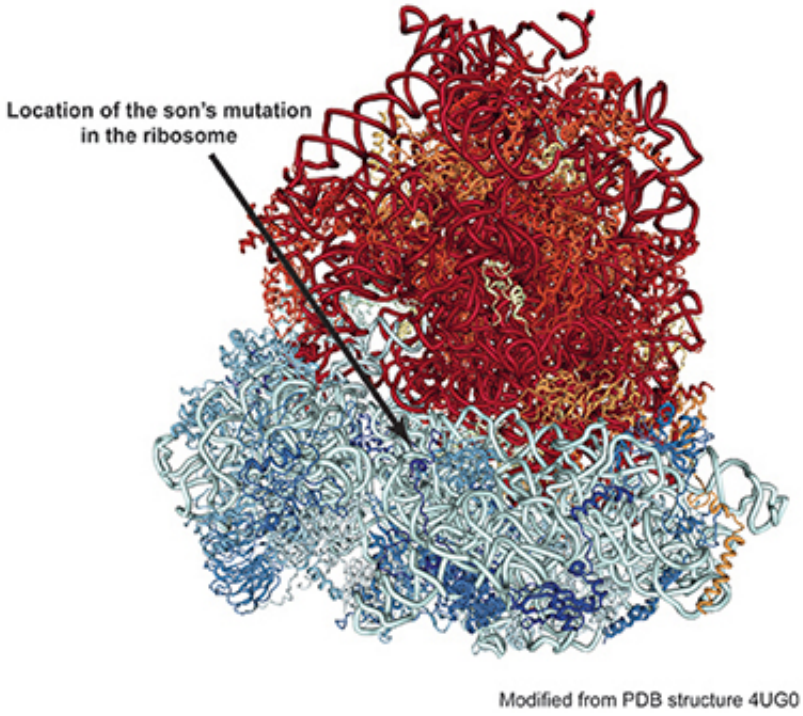

Credit: Yale University

The severe learning disabilities of a teenager in the Netherlands were caused by a genetic mutation that affects the function of ribosomes, the cellular protein-making factories crucial to all life. The new finding by an international collaboration of scientists adds to list of newly identified diseases called ribosomopathies.

The research began in 2013 when Marc Pieterse contacted Susan Baserga, an expert on ribosomes, about the condition of his son, Vincent. Baserga is professor of molecular biophysics and biochemistry, genetics and therapeutic radiology at Yale.

Vincent, then 10, suffers from a form of microcephaly, severe learning disabilities, unusual facial characteristics, and some hearing loss. Geneticists sequenced his genome and found a mutation in the ribosomal protein uS12. However, it was unclear whether the mutation played a role in Vincent's condition.

An M.D./Ph.D. candidate in Baserga's lab, Sam Sondalle, found that the mutation indeed reduced protein function in yeast. Subsequent work with researchers in the Netherlands, the United Kingdom, France, and the University of Maryland helped confirm and extend the findings that the mutation in uS12 caused birth defects in young Pieterse and similar abnormalities in another individual. The findings were published in March 2 in the *American Journal of Human Genetics*.

"I hope in the long run many patients will benefit from this work," said Marc Pieterse.

Provided by Yale University