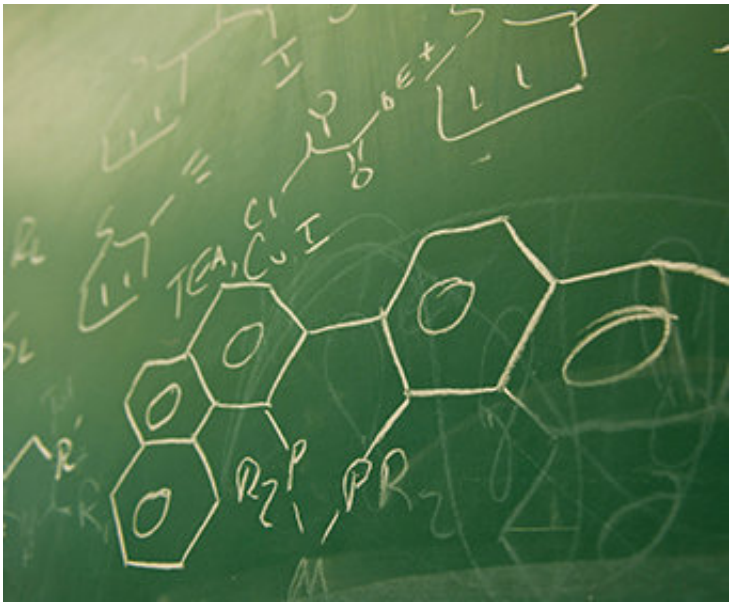
Geneticists explore 30-year old medical case

November 2 2017, by Regina Unger



Credit: University of Texas at San Antonio

Aimin Liu, Lutcher Brown Distinguished Chair in Biochemistry at The University of Texas at San Antonio (UTSA), has published research demystifying a decades-old medical case. The article, which appeared in *Molecular Genetics and Metabolism*, explores the first confirmed human case of 2,3-dioxygenase deficiency.

"Dr. Liu's study is an important step forward in understanding hypertryptophanemia and is sure to have a tremendous positive human impact," said George Perry, dean of the UTSA College of Sciences and Semmes Foundation Endowed University Chair in Neurobiology. "His

work is a vital part of our brain health research efforts and an example of the exciting, innovative research UTSA is known for."

Liu, a UTSA biochemist, specializes in metabolism, the body's process to convert food to energy. His expertise also includes biosynthesis, enzymology and protein biochemistry.

The UTSA researcher collaborated with Patrick Ferreira, a medical geneticist at Alberta Children's Hospital in Calgary, Alberta, Canada, to document the case. Ferreira contacted Liu in 2015 seeking a better understanding of metabolic disorders he had followed in a female baby born in 1986.

Using advanced instrumentation techniques in his UTSA laboratory, Liu concluded that tryptophan 2,3-dioxygenase deficiency was the explanation for the patient's conditions: chronic hypertryptophanemia, characterized by an excess of tryptophan, and its subsequent hyperserotonemia, a condition caused by excessive serotonin. Both disorders are rarely reported in the medical literature.

Their case is now the first confirmed report of human tryptophan 2,3-dioxygenase deficiency.

Tryptophan, an essential amino acid, is necessary for normal growth and is a precursor to the neurotransmitters serotonin and melatonin. Serotonin plays a critical role in the central nervous system and controls many of the body's functions. Melatonin is derived from serotonin and has a critical role in the body's sleeping cycles.

"We are dedicated to conducting this intense research because of the impact it can have on people struggling with these conditions," he said. "The advanced labs we have at UTSA combined with our devoted students is what make these types of discoveries happen."

Liu hopes that the discoveries he, Ferreira and his UTSA students made will lead to the development of medications that could help people with metabolic disorders like the one they studied.

"It was very moving to be involved in a study that could result in a person, or multiple people, benefitting from our work," Liu said. "It's a wonderful example of laboratory work making a difference in the everyday world."

Provided by University of Texas at San Antonio

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