

# ATRX -- Too much or too little underlies sex abnormalities

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6% of the patient population in Melbourne carries a genetic abnormality implicated in thalassemia. As well as causing blood disorders and severe mental retardation, boys with ATRX mutations have genital abnormalities.

Our understanding of the genetic mutations which underlie this cluster of disorders has been advanced significantly by research performed by Dr Anthony Argentaro and just published in the prestigious journal, *Proceedings of the National Academy of Sciences*.

A Melbournian, Dr. Argentaro has solved a major piece of the puzzle about how ATRX causes this wide spectrum of disorders whilst working in Oxford and Cambridge Universities (UK) on an Australian Fellowship.

Employing nuclear magnetic resonance, he solved the three-dimensional structure of part of the huge ATRX protein that carries most of the clinical mutations. By comparing the structure of normal and mutated ATRX, he determined that too much OR too little ATRX leads to developmental disorders.

He has returned to Melbourne to Prince Henry's Institute, where he will utilise his expertise to better understand how ATRX causes abnormalities in sexual development. He will work with international authority, Associate Professor Vincent Harley, to identify the genes and other proteins regulated by ATRX. In doing so, Dr Argentaro aims to

identify targets for treating these disabling and traumatic diseases.

Link: <http://www.pnas.org/cgi/content/full/0704057104>

Source: Research Australia

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