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Researcher offers hope for male diabetes sufferers

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(PhysOrg.com) -- Scientists at Glasgow have made a significant discovery in the study of a rare form of diabetes which predominantly affects men.

Hereditary Diabetes Insipidus is, according to experts, the most common genetic disease known in medicine. It causes serious kidney malfunction and can, if untreated, be fatal. There are an estimated to be around 9000 Heredirary DI patients in the UK.

Although the genetic basis of the disease has been established for a number of years, researchers in the faculty of Biomedical and Life Sciences at Glasgow have now pin-pointed an approach to treatment that may be expected to pave the way for future drug development.

The report has been published in the highly respected international journal, *Proceedings of the National Association of Sciences of the USA (PNAS)*.

Graeme Milligan, Professor of Molecular Pharmacology at the University of Glasgow, and one of the authors on the report, said their findings are of "immense significance" because current strategies used for treatment have limited effect.

He explained: "The gene that is responsible for Hereditary Diabetes Insipidus is found on the X chromosome, which is why it is a predominantly male condition. Genetic defects cause the V2 vasopressin

receptor found in the kidney to malfunction.

"In turn this reduces the capacity of the organ to concentrate urine, which is why patients need to go to the toilet frequently and drink large volumes of water, because the receptor is not responding to Vasopressin or Antidiuretic Hormone (ADH) produced to regulate the body's retention of water. The result of these genetic mutations in the receptor protein mean that it fails to mature, fold and function properly."

"These findings are important for sufferers long term, because our studies have employed novel, early stage drugs that can activate the mutated forms of the V2 vasopressin receptor. Further improvements to these drugs will result in their use by patients."

The work was also carried out with Joris Robben who now works at the University of Nijmegen, in Holland, from Glasgow.

Hereditary Diabetes Insipidus is not the only genetic disease in which mutation of a receptor results in it's inability to fold and function correctly, so the approach we have taken may be equally applicable to these other conditions.

To view the paper abstract please see: www.pnas.org/content/early/200… /0900130106.abstract .

Provided by University of Glasgow