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Scientists discover gene which causes rare disease in babies

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A rare disease which often first presents in newborn babies has been traced to a novel genetic defect, scientists at Queen Mary, University of London have found.

The research, published online in Nature Genetics (27 May) discovered 20 distinct mutations in a specific gene found in patients with the rare adrenal disease, Familial Glucocorticoid Deficiency (FGD).

The potentially fatal disease means affected children are unable to produce a hormone called cortisol which is essential for the body to cope with stress.

Lead researcher Dr Lou Metherell*, endocrine geneticist at Queen Mary, University of London, said: "People who inherit this disease are unable to cope with physical stress. For example, the normal response to infection or traumatic injury is to produce cortisol supporting the metabolic response to the event. Patients with FGD cannot do this and may die if untreated.

"We found 20 distinct defects in the antioxidant gene nicotinamide nucleotide transhydogenase (NNT) in patients from all over the world who suffer from FGD."

The researchers, which include Eirini Meimaridou and Professor Adrian Clark, also at Queen Mary in the William Harvey Research Institute, had previously found defects in four genes present in this disease. The new

research uncovered mutations in NNT, an antioxidant gene, which provides a new mechanism for this adrenal disease.

"Patients with this form of FGD exhibit oxidative stress (OS) in the adrenal, a process which is involved in other diseases such as neurodegenerative conditions, cancer, stroke, diabetes and cardiac dysfunction," Professor Clark said.

"If we can discover how the OS causes its effect then this might give us clues to the mechanism in other diseases like those listed above and it may then be possible to use appropriate drugs to reduce or prevent it."

More information: "Mutations in NNT encoding nicotinamide nucleotide transhydrogenase cause familial glucocorticoid deficiency" was published online in *Nature Genetics* on 27 May 2012.

Provided by Queen Mary, University of London