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Breakthrough in birth-defect research

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Scientists have discovered how to prevent certain craniofacial disorders in what could ultimately lead to at-risk babies being treated in the womb.

University of Manchester researchers, working with colleagues at the Stowers Institute for Medical Research in Kansas, have successfully treated mice with Treacher Collins syndrome – a rare genetic disorder characterised by underdeveloped facial bones, absent or deformed ears and occasionally cleft palate.

The team had previously found that the condition, which affects one in 10,000 individuals, was caused by a mutation in a single gene called TCOF1. They later discovered that this mutation causes cells, known as neural crest cells, to die prematurely in the early stages of pregnancy resulting in the facial anomalies.

Now, writing in the journal *Nature Medicine*, the researchers have shown that preventing the neural crest cells from dying allowed mice with the Treacher Collins gene to develop normally. The principle, say the authors, could also be applied to other single-gene birth defects.

“This is the first time that a congenital defect has been successfully treated and provides genuine hope within a realistic timeframe of one day preventing these conditions in humans,” said Professor Mike Dixon in Manchester’s Faculty of Life Sciences.

“The method we used to stop the cells dying had significant side-effects but there are other ways to prevent cell death and we are confident the

next stage of our research will identify some safe methods."

The anomalies caused by Treacher Collins syndrome, including underdeveloped jaw and cheek bones, occur during the first few weeks of pregnancy.

Since tests to identify the disorder in the unborn child can only be carried out at nine weeks, long after the damage has been done, any future treatment would have to target those babies most at risk.

"Treacher Collins is an inherited disorder, so the hope is we could use this method to prevent parents with the condition passing it on to their children," said Professor Dixon.

"This is an exciting step in our investigations and, once we have found a safe method of stimulating the production of neural crest cells in mice, we can look at early clinical trials in humans."

Source: University of Manchester