

# World first discovery gets to the heart of birth defects

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A frontal section of a mouse heart at embryonic day 17.5 showing normal anatomy, which can be disrupted by short-term exposure to hypoxia during gestation. The image was generated using optical projection tomography. Credit: Company of Biologists

For the first time, scientists believe they've discovered a cause of multiple types of birth defects triggered by environmental stresses.

The breakthrough made by scientists at the Victor Chang Institute, shows that cellular stress could be the key to understanding why many babies are born with defects of the [heart](#), vertebrae and kidney, among others.

Affecting 1 in 100 babies, childhood heart disease is the most common form of birth defect in the world. But despite its prevalence, surprisingly the genetic and environmental causes are very poorly understood.

The research, led by world renowned professor Sally Dunwoodie, analysed the effects of short term oxygen deficiency on heart development in an embryo.

"We obviously know that smoking is terrible for an unborn baby's health. But oxygen deficiency in an embryo can be caused by many things, for example prescription medications, [high blood pressure](#), high altitude, a tangled umbilical cord, as well as carbon monoxide," Professor Dunwoodie explained.

Using a mouse model, the scientists reduced oxygen levels inside a chamber from the normal level of 21 percent to as low as 5.5 percent, for eight hours.

The scientists showed for the first time that reduced oxygen levels damaged the developing heart. The types of heart defects were the same as those most commonly found in humans. Crucially the scientists worked out exactly how the low oxygen was damaging the developing heart.

"We discovered that reduced oxygen triggered a [stress response](#) in the embryonic cells. The cells try to relieve the stress by stopping protein production. Suddenly those proteins aren't available to make the heart at a critical time and the heart couldn't develop properly," Professor Dunwoodie revealed.

Importantly, [oxygen deficiency](#) isn't the only trigger of this cellular stress. There are multiple factors which can set it off, such as a viral infections, increased temperature, high blood glucose, poor nutrition, and pollution.

"This [cellular stress response](#) could be the key to a variety of birth defects, not just [heart defects](#). Now, we strongly suspect it's an underlying mechanism for many different types of [birth defects](#), including those of the vertebrae, kidney and others."

"Surprisingly this [cellular stress](#) response has been used for hundreds of millions of years and it is only now that we have discovered that it can cause organs, such as the heart, not to form properly" added Professor Dunwoodie.

The study has recently been accepted for publication in the journal *Development*.

**More information:** Hongjun Shi et al, Gestational stress induces the unfolded protein response, resulting in heart defects, *Development* (2016). [DOI: 10.1242/dev.136820](https://doi.org/10.1242/dev.136820)

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