

Prof Sally Dunwoodie: For the vast majority of birth defects, we don't know why they occur, and people when they have a child born with a defect they want to know why did this happen, what did I do wrong and is it likely to occur again in my family. So what we've found is twofold, we've identified gene changes and mutations that arise in embryos that can cause the vertebrae to form abnormally and these abnormal vertebrae can lead to scoliosis or bending of the spine. So this is an important finding and it is a world first, so now we have a genetic handle if you like on why multiple defects can develop. And what we've also found, which is significant, is that not everyone with these genetic changes would indeed have a child with a vertebral defect. So the question is why do some embryos develop vertebral defects and others not? And so we reasoned that there's another factor at play here, and we investigated the effect of an environmental factor interacting with a genetic risk factor. And the environmental factor that we looked at was low oxygen or hypoxia. So we were able to show that low oxygen during gestation for a very short period of time could actually increase the likelihood of an embryo that had a genetic risk for a vertebral defect to actually develop a vertebral defect and end up with scoliosis. So the implication of this research is that our findings mean that other research scientists including ourselves can continue along on this route to look at how nature and nurture interact and cause birth defects. For women today, I guess that we just have to be mindful that we play a big role in the development of our embryos and then there might be factors out there that we don't even know are risky so I would suggest that perhaps women need to be mindful of the fact that the environment is important and for women to think about when they're planning on a pregnancy or in the early stages of a pregnancy they're careful of some activities. So for example, prescription drugs can actually cause hypoxia in the embryo and we're talking about a very short, sharp period of time of this environmental insult that can affect embryonic development.

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