

First semi-identical twins identified in pregnancy

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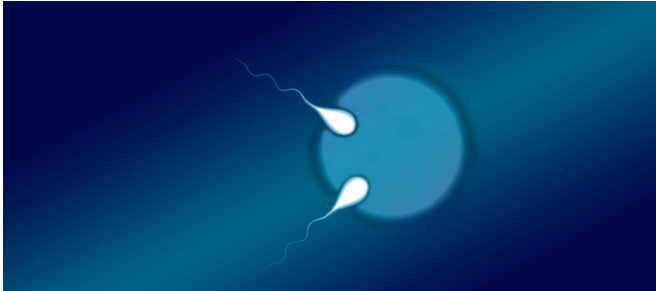


Illustration shows two sperm fertilizing an egg. Credit: QUT

Young Brisbane twins, a boy and a girl, have been identified as only the second set of semi-identical, or sesquizygotic, twins in the world—and the first to be identified by doctors during pregnancy.

The now four-year-old boy and girl are identical (monozygotic) on their mother's side sharing 100 per cent of their mother's DNA, but are like siblings on their father's side, sharing only a proportion of their father's DNA.

The case, the first worldwide to identify semi-[identical twins](#) on [genetic testing](#) while in the womb, has been reported today in The New England Journal of Medicine (NEJM) by fetal medicine specialist and Deputy Vice-Chancellor (Research) at UNSW Professor Nicholas Fisk and Queensland University of Technology (QUT) clinical geneticist and Diagnostic Genomics course coordinator Dr. Michael Gabbett.

Sesquizygotic represents a third type of 'twinning' between identical and fraternal (dizygotic).

"It is likely the mother's egg was fertilised simultaneously by two of the father's sperm before dividing," said Professor Fisk, who led the fetal medicine team that cared for the mother and twins

while based at Royal Brisbane and Women's Hospital in 2014. Professor Fisk, a past President of the International Fetal Medicine and Surgery Society, worked alongside Dr. Gabbett.

"The mother's ultrasound at six weeks showed a single placenta and positioning of amniotic sacs that indicated she was expecting identical twins. However, an ultrasound at 14 weeks showed the twins were male and female, which is not possible for identical twins."

Identical twins result when cells from a single egg fertilised by a single sperm divide into two, so identical twins are the same gender and share identical DNA. Fraternal twins occur when each twin develops from a separate egg and the egg is fertilised by its own sperm.

Dr. Gabbett said if one egg is fertilised by two sperm it results in three sets of chromosomes, one from the mother and two from the father.

"Three sets of chromosomes are typically incompatible with life and embryos do not usually survive," he said.

"In the case of the Brisbane sesquizygotic twins, the fertilised egg appears to have equally divided up the three sets of chromosomes into groups of cells which then split into two, creating the twins.

"Some of the cells contain the chromosomes from the first sperm while the remaining cells contain chromosomes from the second [sperm](#), resulting in the twins sharing only a proportion rather 100 per cent of the same paternal DNA."

Sesquizygotic twins were first reported in the US in 2007. Those twins came to doctors' attention in infancy after one was identified with ambiguous genitalia. On investigation of mixed [chromosomes](#), doctors found the boy and girl were identical on their mother's side but shared around half of their

paternal DNA.

Professor Fisk said an analysis of worldwide twin databases pointed to just how rare sesquizygotic twins are.

"We at first questioned whether there were perhaps other cases which had been wrongly classified or not reported, so examined [genetic data](#) from 968 [fraternal twins](#) and their parents," he said.

"However we found no other sesquizygotic twins in these data, nor any case of semi-identical twins in large global twin studies.

"We know this is an exceptional case of semi-identical twins. While doctors may keep this in mind in apparently identical twins, its rarity means there is no case for routine genetic testing."

The paper, Molecular Support for Heterogonesis Resulting in Sesquizygotic Twinning, is published in *The New England Journal of Medicine* on February 28.

More information: *New England Journal of Medicine* (2019). [DOI: 10.1056/NEJMoa1701313](https://doi.org/10.1056/NEJMoa1701313)

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