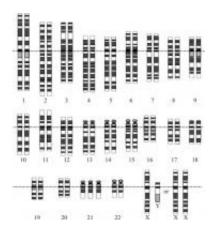


Babies with Down syndrome have healthier hearts

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Karyotype for trisomy Down syndrome. Notice the three copies of chromosome 21. Courtesy: National Human Genome Research Institute

Compared with the early 1990s, babies born with Down syndrome (trisomy 21) have a lower risk of developing serous heart problems, according to a new registry study from Karolinska Institutet published in *Pediatrics*, the journal of the American Academy of Pediatrics. The reason for this improvement is not known, but the researchers believe there to be a likely connection with fetal diagnostics.

To perform their study, the researchers accessed the national Medical Birth Registry, the Birth Defects Registry and the Patient Registry. Amongst the 2.1 million babies born between 1992 and 2012 were almost 2,600 with Down syndrome that were further studied with regard



to congenital heart defects.

The risk dropped

Even though half the babies with Down syndrome had some sort of <u>congenital heart defect</u>, the researchers found that the risk of complicated heart defects dropped sharply during the study period.

"The risk of complicated heart defects that usually require surgery, was 40 per cent lower at the end of the study period," says the project's initiator Stefan Johansson, consultant at Sachs' Children and Youth Hospital and researcher at Karolinska Institutet's Department of Medicine in Solna. "We also observed a shift in incidence ratios, with the decrease in complicated heart defects being matched by an increase in less severe heart defects."

While the project was not designed to analyse causal relations, the group speculates that the result might be attributable to fetal diagnostics. If routine ultrasound reveals a heart defect, the parents usually opt to have the fetus checked for chromosome abnormalities. If Down syndrome is then diagnosed, it is not uncommon for the pregnancy to be terminated, which could account for the drop in the number of babies born with severe heart defects.

Selection effect

Another possibility is that prenatal tests for Down syndrome, which are usually offered to older women, lead to a selection effect if the age of the mother impacts on the risk of <u>heart</u> defect in the fetus.

"The fact that babies born with Down syndrome have healthier hearts than they used to is good news for the families, many of whom find it



very tough at first after the baby is born," says Dr Johansson. "I also hope that our research makes a positive contribution to the public conversation on Down syndrome, which centres mainly on fetal diagnostics. I agree with many parents of these children that the discussion is too tainted by the abortion issue, as if people with Down syndrome don't have the same right to life as others. Which of course they do!"

It is unknown whether today's generation of children with Down syndrome are healthier in other respects too. The research group plans to examine if other common diseases associated with the condition, such as infection sensitivity and hypothyroidism, have also become less common.

More information: S. Bergström et al. Trends in Congenital Heart Defects in Infants With Down Syndrome, *PEDIATRICS* (2016). DOI: 10.1542/peds.2016-0123

Provided by Karolinska Institutet

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