

One in 20 may carry genetic disaster risk

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A genetic screening program for would-be parents has uncovered hundreds in danger of having children with heartbreaking diseases — most of whom had no idea of the risk. Melbourne researchers have found the threat to pregnancies from the three inherited conditions is equivalent to the risk of Down syndrome. The disorders are the incurable lung disease cystic fibrosis, the intellectual disorder fragile X syndrome, and a deadly muscle-wasting condition called spinal muscular atrophy.

While prenatal checks for Down syndrome are routine, screening for the three inherited diseases is usually limited to those with family histories.

The study analysed the first five years of the tests, offered by Victorian Clinical Genetics Services, a non-profit subsidiary of the Murdoch Childrens Research Institute. Of 12,000 people screened — 88 per cent with no previous association with the diseases — about one in 20 turned out to be carriers. The results have been published in the journal *Genetics in Medicine*.

Senior author David Amor said that unlike Down syndrome, which was not caused by inherited mutations, the diseases were potentially predictable. “There’s a tendency to think of these conditions as too rare to warrant this type of testing,” he said.

The average life expectancy of Australians with cystic fibrosis is 38, while few people with spinal muscular atrophy survive beyond infancy. As “autosomal recessive” conditions caused by identical genetic mutations in both parents, the illnesses can skip generations.

Professor Amor said there was a strong argument for screening the wider population for the diseases. Carriers could be detected before they conceived and take steps — through IVF, for example — to guard against having children with the condition.

Melbourne couple Aaron and Kirsty McConnell had no idea each carried the SMA gene when they married. Each had a healthy child already, and new daughter Lily “was passing all her milestones” at four months.

At six months, Lily started struggling to hold herself upright. It was the start of a “very brutal” deterioration of her muscles that affected her ability to eat, sleep, move and ultimately breathe, claiming her life a year later.

“We just happened to be two carriers in the community, and we’ve lost our child because of it,” Ms McConnell said. “Not once did we ever consider that by coming together and having a baby of our own, we would pass on a terminal genetic condition. “There’s a one in four chance that two carriers who have this gene pass it on to their child,” she said. “Those odds are just too high. I wouldn’t want anyone else to experience what we have been through.”

The new study has emerged as rapid improvements in genetics and genomics spawn calls for wider application of the techniques. Professor Amor said that while the tests cost \$385 each, they could deliver substantial health savings because the lifetime expenses of caring for people with the diseases ran into millions of dollars per patient.

“The health economics for genetics and genomics is clouded by the fact that things are changing so fast,” he said. “It takes a long time to do a proper health economic analysis, and suddenly all your figures are out of date.”