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Title:

THE IMPACT OF A NATIONAL POPULATION CARRIER SCREENING PROGRAM ON CYSTIC FIBROSIS BIRTH RATE AND AGE AT DIAGNOSIS: IMPLICATIONS FOR NEWBORN SCREENING

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What was your research question?

The Israeli Ministry of Health has been running a CF population carrier screening (PCS) program since 1999. This is geared at couples planning to conceive and aims to detect healthy carriers of one of the common mutations. We sought to assess if this program resulted in reduction of CF births and earlier diagnosis of those born.

Why is this important?

Carrier screening programs are deemed successful when they capture most mutations and are taken up by a large proportion of the target population. Their aim is to put couples who are found to be carriers in a position to make informed decisions about the continuation of a pregnancy. This includes diagnosing fertilized eggs via in vitro fertilization (IVF) and selecting only unaffected fetuses for implantation. As a result, less children with CF would be born and those born would be expected to carry milder mutations. If a decision is taken to continue the

pregnancy, parents would know about the CF diagnosis from day one, in effect rendering newborn screening superfluous.

What did you do?

We gathered information from a number of sources, including the Central Bureau of Statistics, Ministry of Health and Israeli CF Registry. We calculated the annual CF birth rate between 1990 and 2011. We analyzed children born with CF between 2004 and 2011 in more detail, looking at their age at diagnosis, mutations and disease severity at the time of diagnosis. We classified individuals according to ethnic groups to assess the effect of ethnicity and religious beliefs on population carrier screen uptake.

What did you find?

The carrier screening program has contributed to a clear reduction in CF live births, from 14.5 per 100.000 in 1990 to 6 per 100.000 in 2011. Uptake was high in the total population. However, of those 95 children born with CF between 2004 and 2011, only 22 (23%) had utilized the screening program and only 68 (72%) had 2 CFTR mutations that would have been picked up on the screening panel. 36% were Arabs and 20% orthodox Jews, compared to 20% and 8% respectively, in the general population. Mean age at diagnosis was 6 months. At diagnosis, 56% had respiratory symptoms, 43% failure to thrive and 20% pseudomonas airway infection.

What does this mean and reasons for caution?

Population carrier screening has an important role in making carrier couples aware of their reproductive risk and informing them about possible options. In Israel, it has resulted in CF birth rate reduction. However, not all segments of the population are currently well served, with low uptake particularly in Arabs and Jewish Orthodox for religious reasons and a large number of missed mutations. As a result, there is a delay in diagnosis, with children diagnosed at a mean age of 6 months, as opposed to 3 weeks in countries with newborn screening programs. This often results in severe, potentially preventable lung disease by the time children are diagnosed.

What's next?

We recommend a more balanced approach employing both population carried screening and newborn screening, these two strategies being complementary rather than mutually exclusive.

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