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Prenatal screening for 22q11.2 deletion syndrome: An opportunity to improve neonatal management for a common cause of congenital cardiac defects

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The 22q11.2 deletion syndrome (22q11.2DS) is associated with a variably expressed complex phenotype that includes cardiac abnormalities that are present in approximately 74% of cases. A high proportion of conotruncal and right aortic arch malformations are attributable to 22q11.2DS. Prenatal prevalence may be as high as one in 1000 fetuses. We developed a non-invasive prenatal screening test for 22q11.2DS and four other clinically significant microdeletion syndromes based on analysis of single-nucleotide polymorphisms in maternal cell-free DNA. In 80,449 pregnancies screened for 22q11.2 deletions, we found 263 (0.39%) at high-risk for fetal deletion and six (one in 13,408) maternal carriers. Pregnancy outcome information was available in 153 pregnancies at high-risk for a fetal deletion of which 24 were true-positive. The testing had a positive predictive value of 15.7%, a false-positive-rate of 0.33%, and the estimated prevalence of 22q11.2DS was 1/1255 in the referral population. Of the 24 true-positives, 21 showed prenatal ultrasound abnormalities including 10 with Tetralogy of Fallot, 3 unspecified cardiac malformations, 1 truncus arteriosus, 1 double outlet right ventricle and 1 VSD. A screening enhancement has reduced the false-positive rate to 0.07% and increased the positive predicative value to 44.2%. Screen-positive pregnancies need to be confirmed by chromosome microarray, either on a prenatal sample (amniotic fluid or chorionic villus) or at birth (blood). Prenatal screening provides an opportunity to reduce adverse sequelae in 22q11.2DS through delivery at a tertiary medical center, the earliest possible management of cardiac, velopharyngeal, and other malformations, seizure management, and additional specialty consultation.

Biography

Benn Peter has completed his undergraduate degree at the University of St Andrews, Scotland, PhD from the University of Birmingham, England, and Post-Doctoral training at the University of Pennsylvania. He also holds a DSc degree from the University of St Andrews. For the past 25 years, he has been at the University of Connecticut Health Center and is currently a Professor Emeritus in the Department of Genetics and Genome Sciences. His clinical responsibilities have included oversight of the cytogenetics and prenatal screening laboratories. His research interests have focused on prenatal testing and diagnosis, most recently involving the introduction of non-invasive prenatal testing.

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