IN THIS ISSUE

Confined placental mosaicism does not usually lead to adverse pregnancy outcomes

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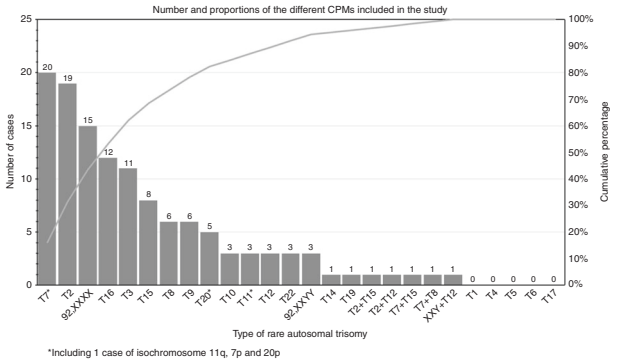

Rare autosomal trisomies (RATs) are associated with adverse pregnancy outcomes; however in 97% of cases, these chromosomal abnormalities exist only in the placenta, not the fetus. This condition, confined placental mosaicism (CPM), has been associated with fetal growth restriction and other pregnancy complications. However, the significance of CPM is controversial due to conflicting studies. In this issue, Grati and colleagues find that only CPM with trisomy 16 (T16) is associated strongly with low birthweight and preterm delivery, suggesting that screening is minimally beneficial. In this cohort study, pregnancy complications in 124 CPM pregnancies were compared with 468 non-CPM matched pregnancies. Chorionic villus samples (CVS) were collected at seven Italian centers for prenatal screening, and then processed in a single laboratory via cytogenetic analysis. If detected by CVS, mosaicism was confirmed by amniocentesis. The RATs most commonly encountered in CPM were trisomies 2, 3, 7, and 16, though many others were also observed. With the exception of T16 and tetraploidies, associations for each individual chromosome abnormality could not be investigated due to small sample size. Thus, all RATs except T16 were analyzed as a group. Although the study only included 12 cases of T16 CPM, a significant association was identified for low birthweight. A weaker association with spontaneous preterm delivery and low Apgar score was also found for T16 CPM. With the exception of T16, no association was found between fetal growth restriction and CPM in this study. Genetic counseling for CPMs based on cell-free DNA testing is difficult because the association with adverse pregnancy outcomes has been unclear. This study shows that in most CPMs involving RATs, excluding T16, the health of the mother and fetus should be comparable to normal pregnancies. The authors conclude that routine detection of CPM might identify 0.8% of those at risk for low birthweight, too low a yield to support screening. *—A. N. Grennell, News Editor*

Genetic services in the US vary widely

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Given the ever-changing health-care environment in the United States and technical advances in genetics and genomics, designs for future genetic services must help close gaps in access to high-quality care. Exactly what priorities and components make a regional system of genetics services effective is unclear. In this report, Kaye and colleagues on the Regional Support Services Model Workgroup and Advisory Committee summarize the current needs identified in stakeholder listening sessions and review the mission and operational model of 14 regional genetic services centers. These data allow genetics services centers to be categorized into eight different models, which vary widely in their missions, strengths, and weaknesses. Based on the priority needs developed in listening sessions and the analysis of existing regional centers, the researchers propose a hybrid model to build a nationwide system that improves access to genetic services. All of the centers touched on a few key themes: regional collaborative structure with a central coordinating office to facilitate resource sharing, using technology to expand availability of health care to underserved populations, and challenges in recruiting a well-trained workforce. However, their highly individualized organizational structures and priorities led to vastly different accessible services, and each of the eight models identified was based on a different priority. In their hybrid model, the research team focuses on improving efficiency of genetic centers through robust technical assistance, supporting nongenetics providers in their care of patients outside the genetics center, and building a regional collaborative structure. Further, integrating with the health-care system by engaging clinicians in identifying and referring patients in need is vital to overcoming systemic barriers. These recommendations are accompanied by two overarching principles to guide future genetic services: family engagement and aligning the goals and activities of regional genetic services support centers. The team concludes that critical gaps in the workforce and in robust data on access to genetic services need to be addressed on a national scale, rather than at regional centers. *—A. N. Grennell, News Editor*

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