18th World Congress in Fetal Medicine

Prediction of facial features of fetal alcohol syndrome at 1 year of age by surface rendered images of the face at 28-31 weeks

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Objective

To explore the possibility of prenatal identification of Fetal Alcohol Syndrome (FAS) by ultrasound biomarkers, to enable earlier intervention.

Methods

Study subjects formed part of the Safe Passage Study (SPS), a large prospective study of approximately 12000 women, including 7000 in Cape Town, South Africa. The main aim of SPS was to establish the contribution of prenatal alcohol exposure (PAE) to stillbirth and sudden infant death. PAE was meticulously documented by means of the time line follow back method. Facial features were assessed by dysmorphologists at 1 year of age and for a positive diagnosis of (partial) FAS at least two facial features were required: palpebral fissure length (measured on a standardised 2D-photograph) < 10th centile of the local reference and a lip and/or philtrum score of 4 or 5 (assessed live). A randomly selected sample (1: 3) of the women underwent additional investigations for ancillary studies. To assess the effect of PAE on fetal face and brain development, 3D ultrasound volumes were obtained at 28-31 weeks' gestation for later offline analysis. A case mix was constructed including 86 cases with a variety of PAE trajectories and 60 unexposed pregnancies. Scoring was done by two or more observers, blinded to exposure and outcome data. Scores were determined on frontal and rotated surface rendered images according to two different lip and philtrum reference guides (Astley for Caucasians and Hoyme for mixed ancestry). The prenatal scores were compared with the lip and philtrum scores at 1 year of age.

Results

Fifteen cases had two or more facial features of (P)FAS at 1 year of age, 4 only had an abnormal philtrum score and 2 only an abnormal lip score. There was a significant correlation between pre- and postnatal philtrum scores (Caucasian reference r=0.3, p=0.0002; mixed race reference r=0.24, p=0.0015) but no correlation for the lip scores. Prenatal philtrum, but not lip, scores were significantly higher in infants meeting (P)FAS facial criteria (p=0.03 Caucasian guide). None of the prenatal scores differed significantly between exposed and non-exposed pregnancies. PAE only affected postnatal philtrum scores (p=0.03) but these were significantly influenced by severity of PAE, smoking and growth deficit at birth.

Conclusion

The prenatal philtrum score at 28-31 weeks is a weak predictor of (P)FAS features at 1 year of age and the postnatal philtrum score. Prenatal lip scores showed no correlation with postnatal features. These findings can be partly due to the limitations of 3D-fetal imaging, but also to significant changes occurring in facial tissues over time.