Prenatal diagnosis of Cri-du-Chat syndrome: five cases

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Introduction
Second-trimester ultrasound abnormalities of Cri-du-Chat syndrome (CDCS) have been well reported. We describe the prenatal presentation of five cases of CDCS.

Methods
We searched the database of two prenatal diagnostic clinics in two public hospitals from July 2010 to February 2018. Chromosome abnormalities were confirmed by IPD followed by karyotyping and aCGH.

Cases
The prevalence was around 1 in 15,000 among all pregnant women. Four patients had TOP for CDCS, and the remaining one had a preterm birth followed by infant death.

Ultrasound abnormalities
Three cases had small cerebellum and prominent lateral ventricles/ cisterna magna detected in the second trimester. One case had absent nasal bone and prominent renal pelvis in the first trimester. However, nil in the other two cases although examination after TOP showed low-set ears in one case and a triangular face in another.

cFTS and cfDNA
In one case, aged 28, positive cFTS with a high-risk for T18 and low PAPP-A. However, cfDNA testing showed a low-risk result for 5p deletion. Amniocentesis showed a 10.14Mb copy loss in 5p15.33-p15.2 on karyotyping and aCGH.
In another case, aged 38, both cFTS and scan showed normal findings. However, cfDNA testing using MPSS at 12 weeks showed reduced amount of DNA in 5p, and the fetal fraction was 12.1%. Amniocentesis followed by karyotyping and aCGH showed a 10.50Mb copy loss in 5p15.33-p15.2.

Conclusion
CDCS is rare in the Chinese population. It can be presented in the first trimester as ultrasound abnormality, low PAPP-A, or reduced amount of cfDNA in 5p in maternal plasma.