Objective
To investigate SLC25A13 gene mutations in 20 families affected by citrin deficiency, as well as the feasibility of prenatal diagnosis of this disease.

Methods
Mutational analysis of SLC25A13 gene was performed by such tools as PCR-RFLP and Sanger sequencing in 20 probands and their parents. For prenatal diagnosis, mothers of the probands undertook amniocentesis of the next pregnancy, and the amniotic fluid cells were cultured for mutational analysis of SLC25A13 gene.

Results
Biallelic pathogenic mutations of SLC25A13 gene were identified in all probands. Among them, two died of hepatic failure and hepatic encephalopathy, respectively, but the remaining 18 had favorable outcomes thus far. A total of 24 fetuses underwent prenatal diagnosis, and 8 had normal genotypes, 11 were mutation carriers while 5 harbored biallelic mutations. Fetuses with wild type or heterozygous SLC25A13 mutations were delivered, and postnatal SLC25A13 analysis proved in accordance with the prenatal findings. In addition, 2 fetuses homozygous for c.851del4 were delivered while 3 pregnancies with fetuses harboring biallelic mutations were terminated.

Conclusion
Mutational analysis of SLC25A13 in families affected by citrin deficiency not only provides laboratory evidences for molecular diagnosis of proband, genetic counseling and the next prenatal diagnosis, but also effectively reduces the risk of recurrence of defective children.