Early prenatal diagnosis of osteogenesis imperfecta by 2D and 3D ultrasound
Chen L, Yao Y, Lin K, Liu K
Chang Bing Show Chwan Memorial Hospital, Changhua, Taiwan

Objective
Osteogenesis imperfecta (OI) is one of the fetal skeletal dysplasia and presenting a heterogeneous group of collagen disorders. Ultrasonography is acknowledged as a reliable diagnostic modality for the prenatal diagnosis of OI, especially type II characterised by ubiquitous and diffuse fractures, thoracic hypoplasia and hypomineralisation of the calvarium. This is a report of detecting OI using 2D B-mode ultrasound and three-dimensional (3D) US.

Methods
A 28-year-old healthy woman (gravida 1, para 0) at 12 weeks gestation for 1st trimester screening and following scanning at 19 weeks.

Results
The ultrasound scan at 12 weeks of gestational age revealed hypoplastic nasal bone, thickened NT and curved legs; in following exam at 19 weeks, 2D ultrasound shows severe micromelia and ubiquitous fractures. 3D ultrasound shows fetal appearance of skull shape abnormality, micrognathia, hypertelorism and low set ear in surface mode. Furthermore, 3D ultrasound in skeleton’s mode delineates clearly the fetal bones’ development (hypomineralization of the calvarium and normal ossification of spine). Compared with 2D US, 3D US can detect fetal OI precisely, and provide additional vivid illustration after various modes of reconstruction that 2D US cannot. After genetic counseling, the patient opted for termination of pregnancy. It was terminated in the 20 week of gestation. Postnatal x-ray also confirmed antenatal findings of ultrasound.

Conclusion
In conclusion, 2D and 3D US have different roles in the diagnosis of lethal skeletal dysplasia. 3D ultrasound provides a novel visual depiction of this defect using in the early prenatal diagnosis of OI is important for pregnancy counseling.