First trimester detection of renal agenesis using 3D ultrasound and Virtual Reality

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Congenital absence of kidneys, also known as renal agenesis occurs in 0.2/1000 births and can be bi- or unilateral in presentation. If bilateral, it is considered lethal; which makes early detection important. Since its etiology is multifactorial, diagnosis warrants a detailed anomaly scan to look for associated anomalies. State-of-the-art ultrasound (US) and visualization techniques such as 3D Virtual Reality (VR) can be used for diagnostics.

Case – 32-year old G2P1 had an ultrasound examination with 3D Virtual Reality at 13+3 weeks’ GA. No fetal bladder and kidneys were visualized. At 13+6 weeks a pelvic kidney and no bladder filling were seen. A 16 weeks exam showed oligohydramnios. Post mortem exam noted an absent right kidney, a multicystic pelvic kidney and ambiguous genitalia.

Introduction

Congenital absence of kidneys, also known as renal agenesis occurs in 0.2/1000 births and can be bi- or unilateral in presentation. If bilateral, it is considered lethal; which makes early detection important. Since its etiology is multifactorial, diagnosis warrants a detailed anomaly scan to look for associated anomalies. State-of-the-art ultrasound (US) and visualization techniques such as 3D Virtual Reality (VR) can be used for diagnostics.

Case

A 32-year old G2P1 visited our center for an advanced US examination. Her first child was known with a complex congenital heart defect, postnatally detected. In this pregnancy the patient decided not to perform first trimester aneuploidy screening.

At 13+3 weeks’ gestational age (GA) an advanced transvaginal first trimester US examination was performed. In addition to regular 2D images, 3D images and VR techniques were used for structural examination. Fetal biometry was normal. We were not able to visualize the fetal bladder and kidneys; using color Doppler US no renal arteries were seen. 3D VR helped to assure both renal fossa were empty. At 13+6 weeks’ GA an echolucent structure was seen in the fetal pelvis, suggestive of a pelvic kidney, again no bladder filling was detected. At 16+0 weeks’ GA US examination revealed there was oligohydramnios. Amniocentesis was performed. QF-PCR and 0.15Mb SNP-array had a normal result. Parents decided to terminate the pregnancy at 17+0 weeks. Post mortem examination is described in Figure 1.

Conclusion

Nonfunctioning kidneys are considered lethal and can be diagnosed in first trimester. 3D VR may aid in the early detection of severe urinary tract anomalies.

Figure 1. A & B: coronal view of 3D Virtual Reality at 13 weeks’ GA in which normal kidneys can be seen. C: The same view at 13+3 weeks’ GA with no visualization of the kidneys. D: post mortem examination with normal adrenal glands ( ), a dysplastic multicystic pelvic kidney ( ), absent right kidney and ambiguous genitalia.

Absent or nonfunctioning kidneys lead to pulmonary hypoplasia and are considered lethal. First trimester detection is possible, as shown in this case. At 13+3 weeks’ GA no fetal kidneys were seen in the renal fossa. Also no bladder filling was observed. 3D VR ultrasound may aid in the early detection of these severe urinary tract anomalies.