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(RESEARCH ARTICLE)

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Bilateral renal agenesis/hypoplasia associated with oligohydramnios and major intrauterine growth restriction: Prenatal ultrasound study

Dinu-Florin Albu [1], Cristina-Crenguța Albu [2,*] and Ștefan-Dimitrie Albu [2]

[1] *"Panait Sîrbu" Hospital, 5 Giulești Street, 6th District, 060251, Bucharest, Romania.*
[2] *"Carol Davila" University of Medicine and Pharmacy, 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.*

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Abstract

Bilateral renal agenesis/hypoplasia/dysplasia is a lethal malformation in humans with an incidence of 1.3 per 10,000 live births. In the etiology of bilateral renal agenesis/hypoplasia/dysplasia, the genetic factor plays an important role. In addition to genetic factors, the etiology of bilateral renal agenesis/hypoplasia/dysplasia also involves the teratogenic effect of hyperglycemia on the embryo in mothers with insulin-dependent maternal diabetes.

The purpose of this paper is to present a special case of bilateral renal agenesis/hypoplasia/dysplasia, which was successfully diagnosed prenatally by ultrasonography, confirmed and managed appropriately, so as to limit the incidence of serious, lethal congenital malformations.

Keywords: Renal agenesis; Renal hypoplasia; Prenatal diagnosis; Ultrasound; Congenital malformation

1. Introduction

Bilateral renal agenesis/hypoplasia/dysplasia (BRAHD) is a lethal malformation in humans [1, 2]. According to EUROCAT, the incidence of BRAHD is 1.3 per 10,000 live births, the condition being 2.5 times more common in men than in women [1, 3, 4].

In the etiology of BRAHD, the genetic factor plays an important role, as evidenced by cases of familial aggregation that suggest an autosomal dominant transmission with incomplete penetrance and variable expressivity [1, 5, 6].

BRAHD can also be a component of some genetic syndromes, such as Fraser syndrome and Branchio-Oto-Renal syndrome [1, 7].

In addition, RET genes are strong candidates for BRAHD, with RET mutations being associated with renal agenesis in mouse embryos [8, 9].

At the same time, a missense mutation of the PAX2 gene was identified in a family with renal dysplasia [10-12].

In addition to genetic factors, the etiology of BRAHD also involves the teratogenic effect of hyperglycemia on the embryo in mothers with insulin-dependent maternal diabetes [13, 14].

*Corresponding author: Cristina-Crenguța Albu
"Carol Davila" University of Medicine and Pharmacy, 37 Dionisie Lupu Street, 1st District, 020021, Bucharest, Romania.

Renal agenesis can be confirmed ultrasonically, with empty renal fossae and absent bladder filling, along with severe oligohydramnios or anhydramnios [15-21].

The purpose of this paper is to present a special case of BRAHD, which was successfully diagnosed prenatally by ultrasonography, confirmed and managed appropriately, so as to limit the incidence of serious, lethal congenital malformations.

2. Material and methods

A 21-year-old Caucasian woman, who was expecting her first child, and who did not have any history of pathological personal antecedents, presented to our Department of Maternal-Fetal Medicine for ultrasound screening of fetal malformations.

In accordance with the internal procedures of the clinic, after the patient had signed an informed consent form and given her consent for the ultrasound investigation, fetal morphology was performed transabdominally using a Voluson E10 Ultrasound device by an experienced obstetrician with qualification in maternal-fetal medicine.

3. Results and discussion

Fetal morphology evaluation highlighted a mono-fetal pregnancy, 21 weeks in evolution, with an estimated weight of 234 grams.

Based on the biometric data, we outlined the following results: well-developed skull, with a structure and morphology within normal limits; nasal bone of 5.5 mm; hard palate apparently integer; oral cavity that seems to have a normal shape of the lips, normoglossia, normognathia and swallowing movements present; nuchal fold thickness 3.1 mm; thorax of normal shape; heart of normal appearance and a fetal heart rate of 149 beats per minute; abdomen of normal size and structure; liver and spleen normally positioned and of normal appearance; antero-posterior abdominal diameter: 36.8 mm; transverse abdominal diameter: 36.9 mm; abdominal circumference: 116.9 mm; ileal loops with normal echogenicity (Fig. 1).

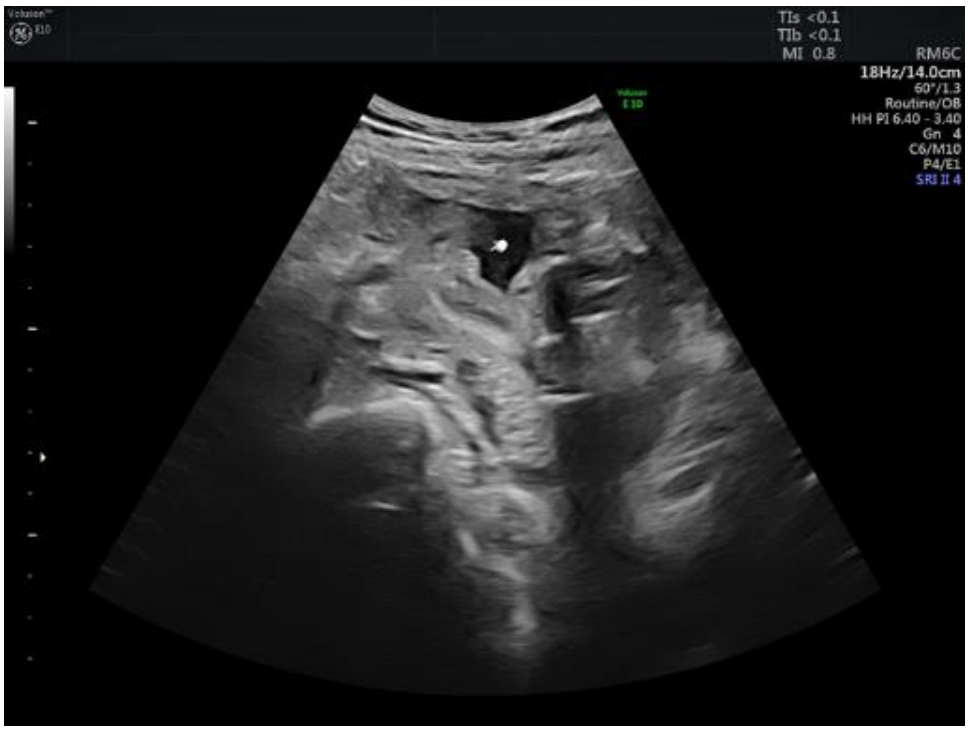

Figure 1 SRI II 4 Ultrasound examination at 21 weeks of gestation showing the nuchal fold thickness

The detailed ultrasound examination of the kidney lodges revealed: right kidney with dimensions: 12.9 / 5.9 mm, with paucivascular-hypoplastic appearance, on Doppler examination, and renal basin of 7.2 mm; left kidney is not evident; urinary bladder visible, in very reduced repletion (Fig. 2 - 4). The fetal sex visualized was female.

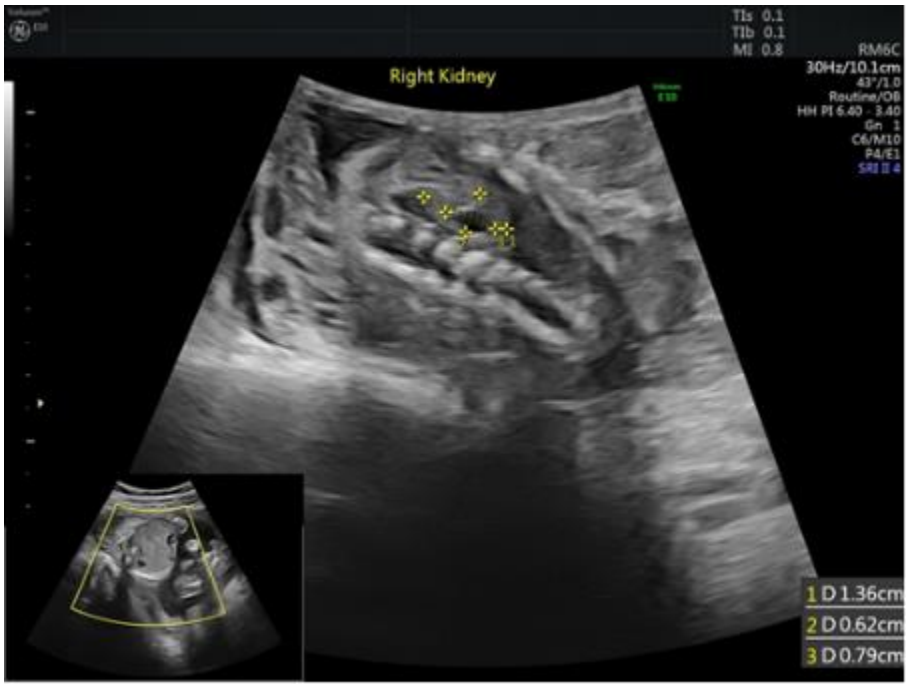

Figure 2 SRI II 4 Ultrasound examination at 21 weeks of gestation showing the right kidney

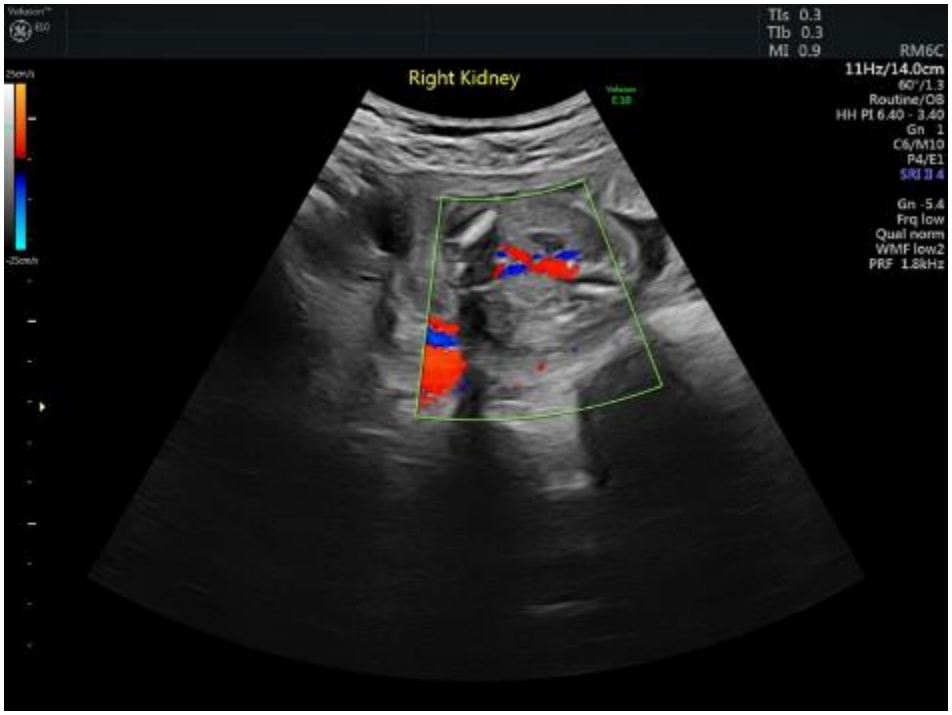

Figure 3 Prenatal images of Color Doppler ultrasonography at 21 weeks of gestation showing the right kidney

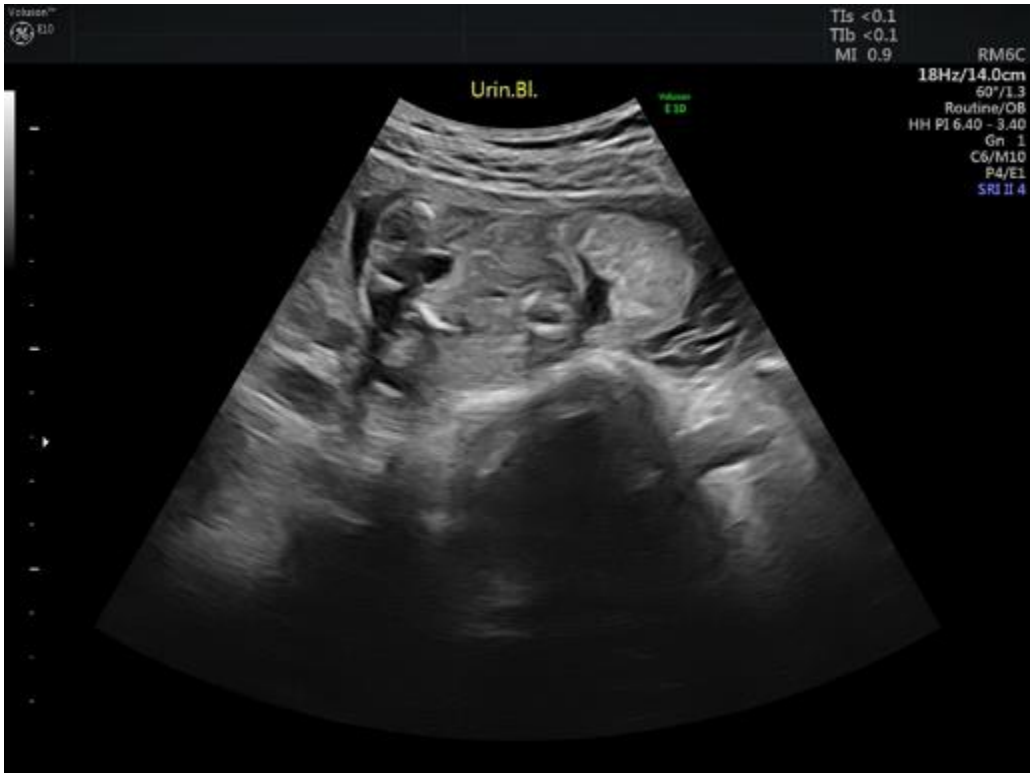

Figure 4 SRI II 4 Ultrasound examination at 21 weeks of gestation showing the urinary bladder

The ultrasound investigation continued with the examination of the extremities which pointed to the following: tri-segmental upper and lower limbs, normally shaped, oligohydramnios, umbilical cord and placenta with regular aspect, without other congenital anomalies visible in the ultrasound examination (Fig. 5 - 7).

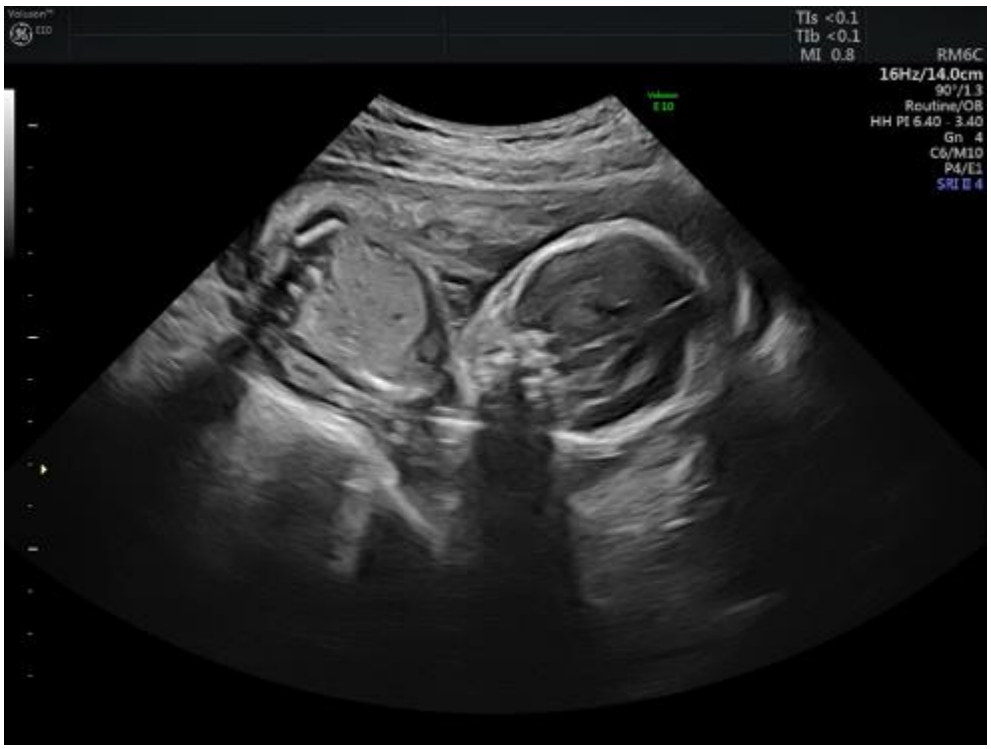

Figure 5 SRI II 4 Ultrasound examination at 21 weeks of gestation showing oligohydramnios

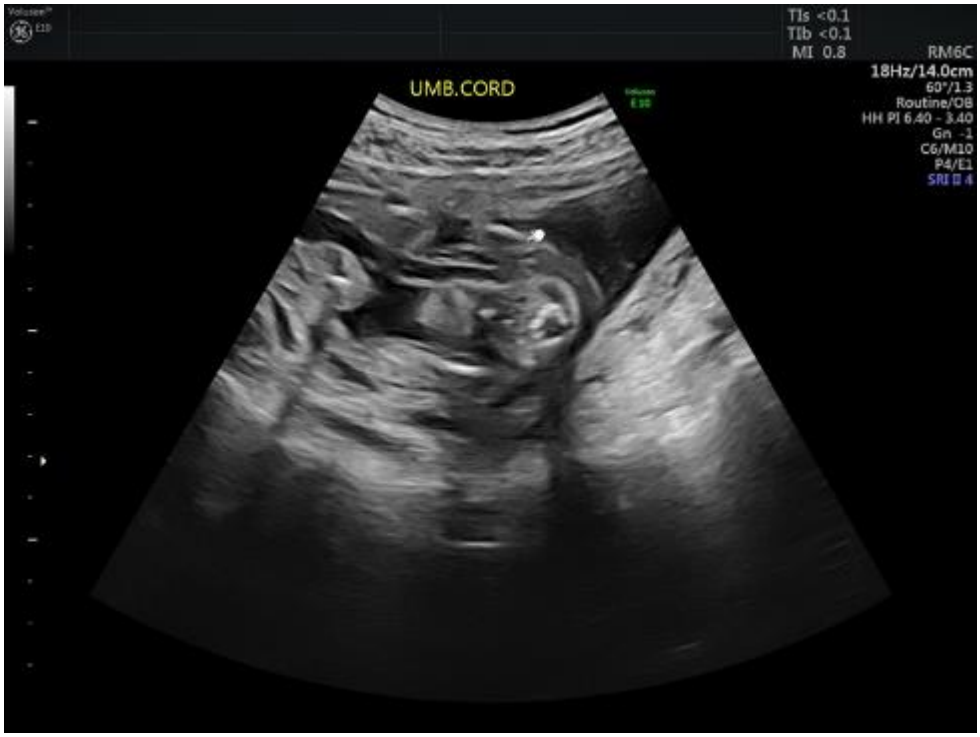

Figure 6 SRI II 4 Ultrasound examination at 21 weeks of gestation showing the umbilical cord

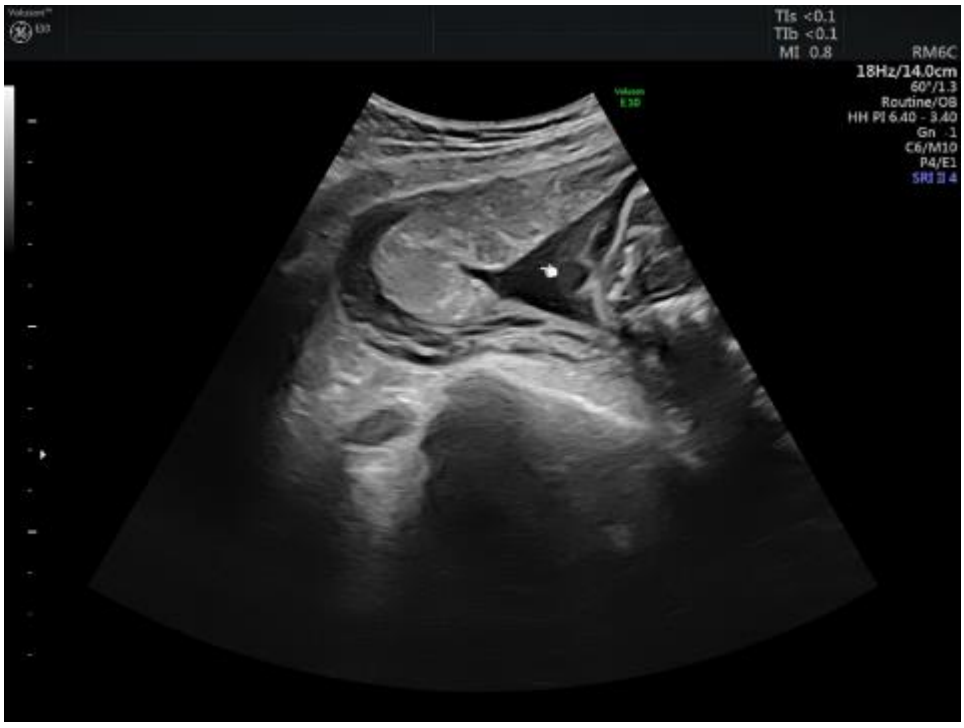

Figure 7 SRI II 4 Ultrasound examination at 21 weeks of gestation showing the placenta

According to the fetal morphology, the following ultrasound diagnosis was established: Mono-fetal pregnancy 21.2 (chronologically) / 18.9 (biometrically) weeks, in evolution; BRAHD: Right renal hypoplasia. Left renal agenesis; Oligohydramnios; Major intrauterine growth restriction (IUGR). Estimated fetal weight: 234 g.

Following the corroboration of all prenatal investigation data, the diagnosis of BRAHD, a lethal congenital malformation, was established, which is why the parents requested the termination of the pregnancy.

4. Conclusion

In conclusion, the presented case is a rare, isolated, spontaneous, non-syndromic case of BRAHD: Right renal hypoplasia. Left renal agenesis, which was successfully diagnosed by prenatal ultrasound examination at 21 weeks of gestation and confirmed postnatally anatomopathological.

Compliance with ethical standards

Acknowledgments

We gratefully acknowledge the patient for giving the necessary permission to report this case.

Disclosure of conflict of interest

The authors declare no conflict of interest.

Statement of informed consent

Informed consent was obtained from the patient included in the study.

Authors' contributions

Authors C.-C.A., D.-F.A. and Ş.-D.A. contributed to this work in conceptualization, methodology, software, and formal analysis. Ş.-D.A.contributed in software, formal analysis, and data curation. C.-C.A. and D.-F.A. contributed in validation, supervision, project administration. All authors read and approved the final version of the manuscript.

References

[1] Harewood L, Liu M, Keeling J, Howatson A, Whiteford M, Branney P, Evans M, Fantes J, Fitzpatrick DR. Bilateral renal agenesis/hypoplasia/dysplasia (BRAHD): postmortem analysis of 45 cases with breakpoint mapping of two de novo translocations. PLoS One. 2010 Aug 25;5(8):e12375. doi: 10.1371/journal.pone.0012375. PMID: 20811621; PMCID: PMC2928268.

[2] Albu CC, Vasilache A, Tanase M, Stanciu IA, Albu DF, Albu SD. The power of noninvasive prenatal diagnosis in the early detection of severe structural birth defects. World Journal of Pharmaceutical Research, 2021; 10(5): 39-46. DOI: 10.20959/wjpr20215-20315. Available from: https://wjpr.s3.ap-south-1.amazonaws.com/article_issue/1619775326.pdf

[3] Renal agenesis/hypoplasia [Internet]. Centers for Disease Control and Prevention. Centers for Disease Control and Prevention; 2021 [cited 2022Nov18]. Available from: https://www.cdc.gov/ncbddd/birthdefects/surveillancemanual/quick-reference-handbook/renal-agenesis-hypoplasia.html

[4] Renal agenesis, Bilateral [Internet]. NORD (National Organization for Rare Disorders). 2021 [cited 2022Nov18]. Available from: https://rarediseases.org/rare-diseases/renal-agenesis-bilateral/

[5] Capone VP, Morello W, Taroni F, Montini G. Genetics of Congenital Anomalies of the Kidney and Urinary Tract: The Current State of Play. Int J Mol Sci. 2017 Apr 11;18(4):796. doi: 10.3390/ijms18040796. PMID: 28398236; PMCID: PMC5412380.

[6] Vivante A, Kohl S, Hwang DY, Dworschak GC, Hildebrandt F. Single-gene causes of congenital anomalies of the kidney and urinary tract (CAKUT) in humans. Pediatr Nephrol. 2014 Apr;29(4):695-704. doi: 10.1007/s00467-013-2684-4. Epub 2014 Jan 8. PMID: 24398540; PMCID: PMC4676405.

[7] Fraser syndrome [Internet]. NORD (National Organization for Rare Disorders). 2020 [cited 2022Nov18]. Available from: https://rarediseases.org/rare-diseases/fraser-syndrome/

[8] Skinner MA, Safford SD, Reeves JG, Jackson ME, Freemerman AJ. Renal aplasia in humans is associated with RET mutations. Am J Hum Genet. 2008 Feb;82(2):344-51. doi: 10.1016/j.ajhg.2007.10.008. Epub 2008 Jan 31. PMID: 18252215; PMCID: PMC2427293.

[9] Costantini F. Genetic controls and cellular behaviors in branching morphogenesis of the renal collecting system. Wiley Interdiscip Rev Dev Biol. 2012 Sep-Oct;1(5):693-713. doi: 10.1002/wdev.52. PMID: 22942910; PMCID: PMC3430146.

[10] Chang YM, Chen CC, Lee NC, Sung JM, Chou YY, Chiou YY. PAX2 Mutation-Related Renal Hypodysplasia: Review of the Literature and Three Case Reports. Front Pediatr. 2022 Jan 11;9:765929. doi: 10.3389/fped.2021.765929. PMID: 35087773; PMCID: PMC8787321.

[11] Nishimoto K, Iijima K, Shirakawa T, Kitagawa K, Satomura K, Nakamura H, Yoshikawa N. PAX2 gene mutation in a family with isolated renal hypoplasia. J Am Soc Nephrol. 2001 Aug;12(8):1769-1772. doi: 10.1681/ASN.V1281769. PMID: 11461952.

[12] Liu S, Zhang P, Wu J, Chang Q. A novel PAX2 heterozygous mutation in a family with Papillorenal syndrome: A case report and review of the literature. Am J Ophthalmol Case Rep. 2021 Apr 22;22:101091. doi: 10.1016/j.ajoc.2021.101091. PMID: 33997468; PMCID: PMC8102412.

[13] Albu CC, Imre M, Tancu AMC, Albu SD, Albu DF, Bencze MA. Diabetic embryopathy or a new genetic syndrome. European Journal of Pharmaceutical and Medical Research. 2021;8(1), 629-663. Available from: https://storage.googleapis.com/journal-uploads/ejpmr/article_issue/1609583064.pdf

[14] Castori M. Diabetic embryopathy: a developmental perspective from fertilization to adulthood. Mol Syndromol. 2013 Feb;4(1-2):74-86. doi: 10.1159/000345205. PMID: 23653578; PMCID: PMC3638774.

[15] Albu CC, Albu DF, Pătraşcu A, Albu ŞD, Efrem IC, Gogănău AM. Prenatal diagnosis of syndromic alobar holoprosencephaly associated with digynic triploidy fetus. Rom J Morphol Embryol. 2020 Oct-Dec;61(4):1309-1316. doi: 10.47162/RJME.61.4.32. PMID: 34171079; PMCID: PMC8343603.

[16] Dias T, Sairam S, Kumarasiri S. Ultrasound diagnosis of fetal renal abnormalities. Best Pract Res Clin Obstet Gynaecol. 2014 Apr;28(3):403-15. doi: 10.1016/j.bpobgyn.2014.01.009. Epub 2014 Jan 29. PMID: 24524801.

[17] Dinu-Florin Albu, Cristina-Crenguta Albu, Stefan-Dimitrie Albu. A Dandy-Walker Variant Prenatally Diagnosed Using Ultrasound on One of the Fetuses of a Twin Pregnancy Obtained through In Vitro Fertilization. Int J Med Res Rev 2015;3(1):127-131. doi:10.17511/ijmrr.2015.i01.022.

[18] Albu CC, Albu DF, Pătraşcu A, Albu ŞD, Efrem IC, Gogănău AM. Prenatal diagnosis of syndromic alobar holoprosencephaly associated with digynic triploidy fetus. Rom J Morphol Embryol. 2020 Oct-Dec;61(4):1309-1316. doi: 10.47162/RJME.61.4.32. PMID: 34171079; PMCID: PMC8343603.

[19] Dinu-Florin Albu, Cristina-Crenguta Albu, Stefan-Dimitrie Albu. The Utility of Antenatal Ultrasound in intrauterine Early Diagnosis of an Autosomal recessive Polycystic Kidney Disease in Fetus. Int J Med Res Rev 2015;3(1):112-114. doi: 10.17511/ijmrr.2015.i1.18

[20] Albu CC, Stancu IG, Grigore LG, Albu DF, Albu ŞD, Pătraşcu A, Gogănău AM. Impact of genetic testing and family health history of cystic fibrosis in the early prenatal diagnosis and prevention of a new case of genetic disorder. Rom J Morphol Embryol. 2019;60(2):667-671. PMID: 31658342.

[21] Albu CC, Albu D, Albu S, Patrascu A, Musat A, Goganau AM. Early Prenatal Diagnosis of an Extremely Rare Association of Down Syndrome and Transposition of the Great Vessels. Rev. Chim.[internet]. 2019 Jul;70(7):2574-2578. Available from: https://doi.org/10.37358/RC.19.7.7383