Survival in a functionally anephric neonate with autosomal recessive polycystic kidney disease (ARPKD)

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Introduction

- Anephric fetus → nonviable
- Is aggressive management an option?
  - Renal agenesis – case reports of survival past the neonatal period [1,2]
- Our patient:
  - Prenatally diagnosed ARPKD and anhydramnios who successfully underwent postnatal nephrectomy and neonatal dialysis as a bridge to kidney transplant

1. George L, Manimtim W, Sharma J. A Singleton Infant with Bilateral Renal Agenesis and Normal Pulmonary Function. Case Rep Pediatr. 2017;2017:1710371.
2. Bienstock JL, Birsner ML, Coleman F, Hueppchen NA. Successful in utero intervention for bilateral renal agenesis. Obstet Gynecol. 2014 Aug;124(2 Pt 2 Suppl 1):413–5.

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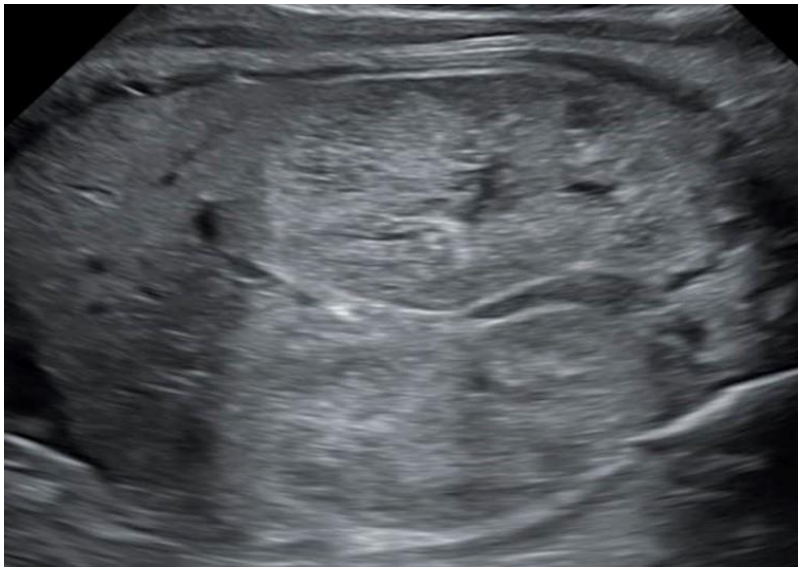
Presentation

- 27y G2P1 presents at 33/40wks after prenatal ultrasound revealed enlarged echogenic kidneys and new anhydramnios consistent with ARPKD



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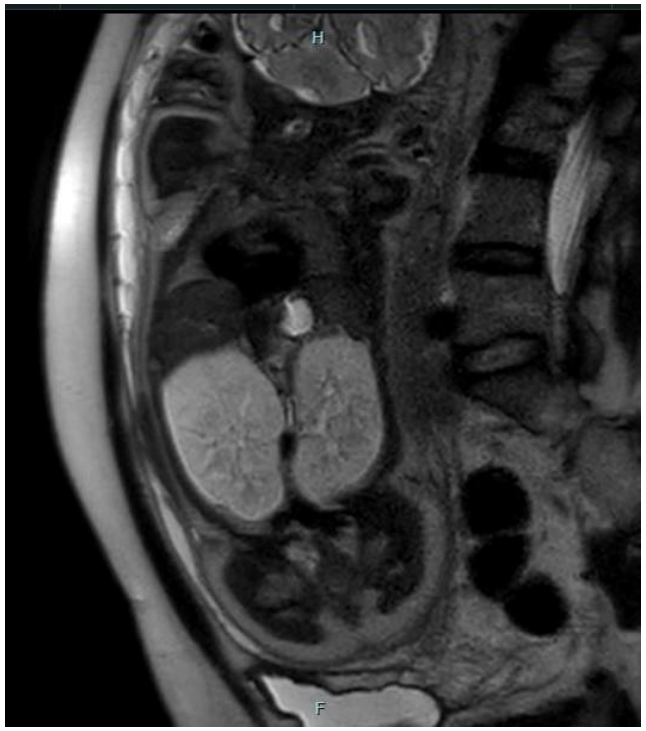
Presentation

- 27y G2P1 presents at 33/40wks after prenatal ultrasound revealed enlarged echogenic kidneys and new anhydramnios consistent with ARPKD
- Fetal MRI confirmed enlarged, hyperintense kidneys and empty bladder.
- Pulmonary survival was deemed likely
- Parents pursue maximal intervention



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Post-natal course - initial

- Born at 36wks
- Anuric as expected
- Left nephrectomy day of life 2 to create space for peritoneal dialysis, with plans for right nephrectomy at time of transplant
- Temporary hemodialysis catheter and peritoneal dialysis catheter placed at this time

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Post-natal course - complications

- Recurrent bacteremia
- Bacterial peritonitis
- Line thrombosis
- Feeding intolerance
- ARDS
- Difficulty uptitrating peritoneal dialysis volumes
  - → Prolonged hemodialysis
- Septic thrombophlebitis with embolic stroke, subsequent hypotonia

- At 1 year, at home tolerating goal enteric feeds on a stable peritoneal dialysis regimen

- Candidacy for transplant?

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2014

"She is a fighter," says Jaime of Abigail (at home with Dan).

MEDICAL MIRACLE

THE GIRL WHO LIVED

Doctors told Rep. Jaime Herrera Beutler her baby would die. But a surprising treatment has made her the first to survive a rare syndrome BY NICOLE WEISENSEE EGAN

Among the family photos in Jaime Herrera Beutler's Camas, Wash., home is an ultrasound, an early image of her daughter Abigail. "I framed it," she explains of the grainy black-and-white square, "because I didn't know how many pictures of her I was going to get."

Shortly before that picture was made, Beutler, a Republican congresswoman, and her husband, Dan, received devastating news: Their unborn child had Potter syndrome, a prenatal condition affecting 1 in 3,000, in which neither the kidneys nor lungs develop. "The doctors said, 'It's 100 percent fatal,'" recalls Jaime. "She will either be stillborn, you'll miscarry, or she will suffocate in your arms after she's born."

Instead Abigail became the first person to survive with Potter's, a milestone that Max Muenke, chief of medical genetics at the National Human Genome Research Institute, calls "a big success—unusual and wonderful."

In her 20th week of pregnancy, however, that outcome was impossible to imagine. A lot of women, her doctor told her and Dan, "would be scheduling a termination now," says Jaime, who opposes abortion. But this wasn't a political vote; it was a deeply personal decision. "As the doctor is saying these things, I could feel her moving," says Jaime, 35. "To me, that was a sign I was not going to be the one that ends this pregnancy." Though she was barely showing, she felt she had to share her story. "I didn't want everybody saying, 'Congratulations!' while inside my heart's breaking."

Beutler at her swearing-in on Jan. 3, 2013.

Photographs by CORAL VON ZUMWALT

Ethics

"Most importantly, ***reports of medical advances belong in the scientific literature – where they can be peer-reviewed by the entire medical community after thorough consideration of all the facts****. Reducing these reports to claims of miracle cures in the lay press may generate desperate parental pleas to try 'anything' masquerading as compassionate care."*

Johnson A, Luks FI. A cautionary note on new fetal interventions. Obstet Gynecol. 2014 Aug;124(2 Pt 2 Suppl 1):411–2.

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Ethics

- Natural history of bilateral renal agenesis:

"With the exception of a single case study using serial amnioinfusion, there has been no other case of survival following dialysis and transplantation documented… based on the ethical analysis of the results from this review, without experimental obstetric intervention, neonatal mortality rates will continue to be 100%. ***Serial amnioinfusion therefore should not be offered as treatment, but only as approved innovation or research.****"*

Thomas AN, McCullough LB, Chervenak FA, Placencia FX. Evidence-based, ethically justified counseling for fetal bilateral renal agenesis. J Perinat Med. 2017 Jul 26;45(5):585–94.

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Moving forward

- Jeremy Sugarman, MD MPH - Bioethicist

- 2018 - in depth ethical review, prompted **Renal Anhydramnios Fetal Therapy (RAFT) trial**

  - Serial amnioinfusion (must begin prior to 26wks GA)
  - 1^{o}: Survival to dialysis
  - 2^{o}: Survival to transplant

https://clinicaltrials.gov/ct2/show/NCT03101891

Sugarman J, Anderson J, Baschat AA, Herrera Beutler J, Bienstock JL, Bunchman TE, et al. Ethical Considerations Concerning Amnioinfusions for Treating Fetal Bilateral Renal Agenesis. Obstetrics & Gynecology. 2018 Jan;131(1):130.

Conclusions

- We report 1 year survival in a functionally anephric patient with ARPKD

- Anephric pulmonary survivors eligible for future kidney transplant *may* be candidates for aggressive perinatal management (serial amnioinfusion, dialysis) in a condition previously considered universally fatal
  - Currently NOT standard of care

- Efforts are underway to further study who is a candidate for such intervention and whether offering such care is ethical

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Thank you

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