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

Presentation

- 27y G2P1 presents at 33/40wks after prenatal ultrasound revealed enlarged echogenic kidneys and new anhydramnios consistent with ARPKD
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• Fetal MRI confirmed enlarged, hyperintense kidneys and empty bladder.

• Pulmonary survival was deemed likely

• Parents pursue maximal intervention
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
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?
Can we?

Should we?
Among the family photos in Jaime Herrera Butler’s home in Washington, D.C., was a ultrasound 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, Butler, 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 Butter. “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—unsual and wonderful.”

In her 20th week of pregnancy, however, that outcome was impossible to imagine. A lot of women, her doctor told her, “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, SS. “To me, that was a sign I wasn’t going to be the one that ended 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.”

**Medical Miracle**

**The Girl Who Lived**

Doctors told Rep. Jaime Herrera Butler her baby would die. But a surprising treatment has made her the first to survive a rare syndrome.

BY NICOLE WEISENSEE EGAN

**Photographs by CORAL VON ZUHWALT**

Children’s Hospital Colorado
“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.”

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.**”

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°: Survival to dialysis
  • 2°: Survival to transplant

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

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
Thank you