A Rare Case of Polycystic Kidney Disease Acquired Through Renal Transplant

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Background

- Autosomal dominant polycystic kidney disease (PCKD) is the most common hereditary disease resulting in end-stage renal disease (ESRD).
- Characterized by development of multiple, bilateral renal cysts with enlargement of the kidneys.

Case Presentation

- CM is a 52-year-old female with ESRD secondary to diabetes mellitus type I who underwent a deceased donor simultaneous pancreas and kidney (SPK) transplant at age 40.
- She presented for her annual transplant evaluation:
  - Was found to have a rise in creatinine to 2.0 mg/dL (baseline 1.4 mg/dL).
  - A new left lower quadrant mass at the site of her renal transplant was noted.
- Ultrasound of the transplant kidney demonstrated enlargement to 19.8 cm with innumerable cysts in the renal parenchyma (largest was 8.6 cm in diameter).
  - Consistent with a diagnosis of PCKD.
- She had no personal or family history of PCKD, and her native kidneys were unaffected.
- The family history of the deceased donor was unknown. However, the recipient of the paired kidney from the same donor also had multiple simple cysts on ultrasound 2 years after transplant.
  - This recipient has since deceased from unknown causes.
- In the five years after the diagnosis of acquired PCKD, CM’s renal function has continued to decline and her current eGFR is 17.
- A recent repeat ultrasound demonstrated further enlargement of her transplant kidney to 30 cm.
- She is now being evaluated for a second renal transplant, potentially from her husband, with transplant nephrectomy at the time of surgery.

Conclusions

- We present a rare case of PCKD acquired via renal transplant from a deceased donor with unknown family history.
- To our knowledge, this is only the third case report of inadvertent transmission of PCKD via transplant.
- In the absence of a family history, a presumptive diagnosis of PCKD can be made with >10 cysts in each kidney.
- Kidney transplant remains the best option for patients with ESRD and it is rare for inherited diseases to be acquired via transplant.
- Our patient’s kidney has functioned for 17 years, and others report 10-15 year survival for transplant of early-stage (pre-clinical) PCKD.
- This argues for a potential role for the transplant of deceased donor kidneys even if the family history includes PCKD.

References