

## Baltimore Polycystic Kidney Disease Center Clinical/Translational Core

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- **Core Resources**

**Study #1:** Longitudinal Cohort Study of adult ADPKD patients without ESRD and eGFR>15 ml/min/1.73m<sup>2</sup>

### **Biorepository:**

- Urine
- Plasma
- Sera
- DNA

### **Clinical Phenotyping**

#### 1. **Clinical history:**

- Age of diagnosis and initial presenting symptoms (e.g., pain, hematuria)
- History of renal and cyst complications: pain, hemorrhage, infection, rupture, stones, hematuria, and nocturia.
- Extra-renal cysts – hepatic, pancreatic, other
- GI complications - diverticulae, herniae (including type and surgical correction)
- Cardiovascular complications - valvular heart disease, aortic aneurysms
- CNS complications - including presence and size of intra-cranial aneurysms
- Exposures - including caffeine, tobacco, iodinated contrast, hormonal therapies and oral contraception, environmental toxins
- Non-ADPKD co-morbid conditions and medications

2. **Family History** – Including age of ESRD of affected family members, and history of extra-renal complications including hepatic disease and intra-cranial aneurysms.

#### 3. **Physical Exam:**

- Resting blood pressure and heart rate
- Height, weight

#### 4. **Clinical Laboratory Results**

- Electrolytes (estimated GFR from CKD-Epi equation)
- Lipid profile,
- Liver function panel,
- Urine protein

5. **Renal and Hepatic MRI:** Total Kidney Volume (TKV) and total liver volume (TLV)

6. **Vascular Function:** Aortic Pulse wave velocity

#### 7. **Echocardiography:**

- Left Ventricular Mass
- Left Ventricular Ejection fraction
- Left ventricular and atrial dimensions
- Valvular dysfunction
- Pericardial effusions

8. **Cognitive Function** (Montreal Cognitive Assessment (MoCA) and Depressive symptoms (Beck Depression Inventory)).

## 9. **Health-related Quality of Life** (NIH PROMIS instruments)

**Study #2:** Human Tissue Study (Adult ADPKD patients with ESRD presenting for simultaneous native nephrectomy and living donor kidney transplant:

### **Clinical Phenotyping:**

1. Demographics
2. Age at onset and presenting symptoms
3. Family History
4. Hypertension
5. Extra-renal Polycystic disease (including liver disease)
6. CNS disease (Intracranial aneurysm)
7. GI complications (including hernias)
8. Blood pressure and treatment
9. Renal ADPKD complications (stones, hematuria, pain, infections)
10. Laboratory data: eGFR (if not on dialysis), blood counts, electrolytes
11. Co-morbid conditions

### **Tissue and biological specimens:**

1. DNA (peripheral blood)
2. Cyst fluid (individual selected cysts)
3. Primary cell cultures from individual selected cysts
4. Tissue blocks