**Title:** “The Impact of Accessible Genetic Testing in an Underserved Pediatric Nephrology Cohort”

Salcedo-Giraldo, J (1). Gaj, K (2). Pitman, T (2). Jandeska, S (2). Zaritsky, J (1,3). Lozano, G (1,3).
1. St. Christopher’s Hospital for Children
2. Natera, Inc.
3. Phoenix Children's Hospital

**Background:** Next generation sequencing (NGS) allows identification of genetic etiologies in a sizable portion of pediatric-onset renal disease patients. Historically, testing has been cost prohibitive which limits access to underserved pediatric populations.

**Objective:** To observe how incorporation of commercially available 385 gene panel for kidney disease impacts clinical diagnosis and management in an underserved pediatric nephrology cohort.

**Design/Methods:** Patients receiving care at St Christopher’s Hospital, Philadelphia PA for kidney disease underwent genetic testing with the RenasightTM test. Positive findings were defined as a single pathogenic (P) or likely pathogenic (LP) variant for an X-linked or autosomal dominant condition, two P/LP variants for an autosomal recessive condition, and the presence of two APOL 1 high-risk alleles (G1 or G2). Other genetic findings that were not considered positives include carriers (one P/LP variant in an autosomal recessive condition) and variants of uncertain significance, which were reported for a subset of patients for whom it was requested.

**Results:** From Oct 2020 to Oct 2021, 194 patients were tested. The median patient age was 14 years. When reported, a high proportion of these individuals identified as African American and/or Hispanic (Table 1) . In addition, 68% of patients were covered by government-based public insurance (Table 2) . Positive findings were identified in 40% (77/194) of patients across 26 genes. The most common positive findings involved the APOL 1 and PKD 1 genes (Figure 1). Positive test results led to changes in medical management in 86% of cases.

**Conclusion:** Genetic testing identified a genetic basis for renal disease in 40% of patients. A large gene panel allows for broad evaluation of multiple genes associated with kidney disease that are otherwise difficult to distinguish clinically. This approach provides accessibility to information that can optimize medical management in underserved patient populations.

**Table 1:** Patient Demographics

**Table 1: Patient Demographics**

| Gender Counts | N | % |
|---|---|---|
| Male | 110 | 57% |
| Female | 84 | 43% |
| **Age Breakdown** | **N** | **%** |
| Age <1 year | 4 | 2% |
| Age 1–<11 years old | 72 | 37% |
| Age 11–<18 years old | 85 | 44% |
| Age 18–21 years old | 16 | 8% |
| Age >21 years old | 17 | 9% |

| Ethnicity counts | N | % |
|---|---|---|
| African American/Black | 35 | 18% |
| Hispanic/Latin American | 35 | 18% |
| European | 14 | 7% |
| Caucasian/Non-Hispanic White | 8 | 4% |
| Asian | 2 | 1% |
| Unknown | 10 | 5% |
| Not Provided | 84 | 43% |
| Other | 6 | 3% |

**Table 2.** Insurance information

**Table 2: Insurance Type**

| Insurance Type | N | % |
|---|---|---|
| Commercial/Private Insurance | 47 | 27% |
| Government Based Insurance | 117 | 68% |
| Other/Unknown | 9 | 5% |

**Figure 1:** Gene variants found through the Renesight Kidney Gene Panel