CRITICAL PATH INSTITUTE

# For Judy Ashley's Family, ADPKD Impact Spans Generations, Sparks Advocacy Work in Community

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Judy Ashley is one of four siblings living with PKD. The family lost their mother when she was just 61, due to complications from dialysis. All five family members and one of Ashley's nieces received transplants. Judy's daughter, who has yet to need a transplant, is also part of a third generation of family members impacted by Autosomal Dominant Polycystic Kidney Disease (ADPKD).

ADPKD is caused by a genetic fault that disrupts the normal functioning of some of the cells in the kidneys causing cysts to grow. Faults in 1 of 2 different genes account for more than 90% of the total cases of ADPKD–PKD1, which accounts for around 78% of cases, and PKD2, which is attributed to around 15% of cases.

"My energy now goes to empowering my daughter to be her own advocate," Ashley explained. "I keep telling her, if it doesn't make sense, if you don't agree with [the treatment plan] keep asking questions."

While keeping her own family informed and confident enough to ask the right questions is the top priority, Judy has also stepped into various roles as an advocate for the public to advance treatments in ADPKD.

Ashley serves as the National Capital Connect Ambassador and as Patient Advocate for the Center of Excellence in Virginia. These roles and organizations bring the PKD community together and provide a forum for patients who want to join in the fight to find treatments and cure for PKD.

"We have meetings and speakers that come and talk about diet, or a nephrologist will advise on what you can expect or what you should look for," Ashley said. "Sometimes we'll have a transplant nephrologist, [mapping out] where someone with PKD is in their journey."

Judy has noticed a disconnect between new patients, and the information they have at their disposal, versus what is out there. C-Path's Polycystic Kidney Disease Outcomes Consortium PKDOC helps to bring the community and key data points together to advance and find new treatments in ADPKD.

PKDOC's drug disease trial models, done in conjunction with the PKD Foundation, academic, industry, and regulatory stakeholders, were used to

support the regulatory qualification of baseline Total Kidney Volume, or TKV, with or without age inclusion. These efforts create a prognostic enrichment biomarker for ADPKD, to predict a 30% decrease in estimated glomerular filtration rate.

PKDOC has successfully qualified TKV as a prognostic enrichment biomarker with both the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA). Furthermore, the FDA designated TKV as a reasonably likely surrogate marker for disease progression in ADPKD. This marker could serve as an endpoint under an accelerated approval pathway followed by a post-marketing confirmation trial showing an effect on the loss of kidney function.

PKDOC also supports its own database that consists of de-identified data from three longitudinal observational patient registries. These data have been standardized and aggregated into a common format using a Clinical Data Interchange Standards Consortium (CDISC) Standard Data Tabulation Model (SDTM) structure. This enables analyses to be performed on a larger expanded dataset. The data covers approximately seven decades of patient visits from the University of Colorado — Denver, Mayo Clinic, and Emory University. In addition, several randomized controlled trial (RCT) datasets (HALT, TAME, ALADIN1) have been ingested.

By creating a unique forum where regulators, industry and academia can interact and share data and insights, the advancement of regulatory-grade solutions can take place. This enables clinical trial optimization and improved decision making to increase late-stage clinical study success rates. As we look towards the near future, PKDOC is well positioned to continue cross-functional multi-stakeholder efforts to advance new prognostic/drug response biomarkers and clinical trial simulators for ADPKD which can directly impact the drug development paradigm in the space.

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