

Abstract Type : Poster

Abstract Submission No. : 1586

Genetic Counseling for Families with Autosomal Dominant Polycystic Kidney Disease (ADPKD) related End-stage Renal Disease (ESRD) : A Case Report

Sang Hyuk Kwak¹, Ju Yen Cho¹, Hyon Ju Kim²

¹Department of Internal Medicine, Kwak Clinic, Korea, Republic of

²Department of Genetic Counseling, Korean Foundation for Rare Disease, Korea, Republic of

Case Study: Case

A fifty-year-old male patient was transferred to our hemodialysis clinic near his residence. In the nephrology department of a university hospital, he was diagnosed with chronic kidney failure caused by ADPKD at the age of forty-seven. Abdominal CT revealed multiple cystic lesions in both kidneys.

According to the patient's family history, he was the fifth of eight siblings, and two of his male siblings had already been diagnosed with chronic renal failure and were getting maintenance hemodialysis. The patient had two adult sons in their thirties and twenties, who were unmarried. The patient feared that his two sons will likewise have end-stage renal failure beyond age of forty. After researching the patient's family history and establishing a family tree, the patient was referred to the Genetic Counseling Clinic at Konyang University Hospital for genetic testing.

Through the Genetic Counseling Clinic's Diagnostic Exome Sequencing Test, a unique likely pathogenic variation(LPV) was identified in the PKD1 gene (c.4221del; p.Phe1408SerfsTer24). The patient was counseled that autosomal dominant inheritance of ADPKD with fifty percent recurrence having ADPKD in high-risk family members, thus genetic testing is also necessary for the patient's children, other siblings, and their progeny.

Conclusion

By the age of sixty, approximately half of patients with ADPKD progressed to end-stage chronic kidney failure. Thus screening, diagnosis, and intervention may be delayed. It is essential for individuals with a family history of ADPKD to undergo comprehensive genetic counseling and genetic testing at a young age, preferably before marriage, and to receive early genetic counseling to prevent transmission of the disease to future generations.