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Fasting in a 16-Year-old Girl at-Risk of Autosomal Dominant Polycystic Kidney Disease

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ABSTRACT

Autosomal dominant polycystic kidney disease (ADPKD) is the most common form of inherited kidney disease that results in renal failure. PKD currently has no causative therapy. However, some treatment options are available, ranging from symptomatic therapy to delaying the onset of end-stage renal failure. Early diagnosis of adult polycystic kidney disease is vital in order to prevent its complications. Ultrasonongraphy and genetic testing are the two preferred diagnostic techniques with defined limitations, mainly regarding age. Herein, we report a case of an ADPKD family whom visited the genetic counseling clinic for determining the disease risk in their symptom-free girls aged 16 and 22 years, and discussing other related issues such as their concern about fasting in Ramadan.

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Introduction

Polycystic kidney disease (PKD) is a genetic disorder inherited as either a dominant or recessive autosomal disease. Autosomal dominant polycystic kidney disease (ADPKD) is the most common form of inherited kidney disease that results in renal failure. Mutations in PKD1 (85%) or PKD2 (15%) genes account for most cases (1). ADPKD caused by mutations in PKD1 gene is significantly more severe with larger kidneys and earlier onset of end-stage renal disease in comparison to PKD2 (2). Loss of function mutations in the mentioned genes promote excessive proliferation of renal tubular epithelia, manifested as cyst formation which increase in number and size with age, besides the formation of intracranial aneurysms (3). Acute and chronic pain and nephrolithiasis are also common complications. Individuals are most commonly affected in midlife, but it may occur from infancy up to the 8th decade of life (4). Each child of an affected individual has a 50% chance of inheriting the mutation and practically, later in all cases with PKD1 or PKD2 mutation, multiple bilateral cysts will develop since the disease penetrance is very high. End-stage renal disease will occur in around 50% of patients by the age of 60 yrs which is the most serious renal complication (5). Overall, 25% of individuals diagnosed with a mutated PKD gene will end up with ESRD and suffer from dialysis. Thus, it is important to perform molecular tests for suspicious ADPKD cases. Moreover, early diagnosis of adult polycystic kidney is vital in order to prevent from its complications. This disorder can be diagnosed through one of the following techniques:

Ultrasound imaging: Begleiter et al. emphasized on the application of ultrasound as a valuable tool in the study of cystic kidney families (6). Sahney et al. suggested that in those suffering from ESRD due to PKD with no previous genetic counseling, any child over 16 years of age should have intravenous pyelography with nephrotomography; and in case of negative study results periodical ultrasonography should be performed up to 25 years of age (7) . However, the clinical use of conventional ultrasonography in ADPKD is currently limited by reduced diagnostic sensitivity, especially in at-risk subjects under the age of 30.

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Genetic testing: Today, for patients suspicious of PKD molecular genetic testing by linkage analysis or direct mutation screening is available clinically (8).

PKD currently has no causative therapy. However, some treatment options are available, ranging from symptomatic therapy to delaying the onset of end-stage renal failure. Such therapeutic options mainly include renal ultrasound examination, abdominal CT or MRI with and without contrast enhancement, standardized blood pressure screening, measurement of blood lipid profile, and urine studies (9).

Herein, we report a case of ADPKD family whom visited the genetic counseling clinic for determining the disease risk in their two children and discussing other related issues.

Case Presentation

A 49-year-old man with confirmed APKD visited the genetic counseling clinic of Iranian Academic Center for Education, Culture and Research (ICECR), Mashhad branch, Iran with his family in May 2014. In a routine medical checkup, the abdominal ultrasonography revealed that he had polycystic kidneys. His family doctor requested an MRI. MRI scanning as a highly sensitive and specific tool for diagnosis of ADPKD ensured the previous findings and thus, he was referred to a clinical geneticist and a genetic counseling center. He had two girls. His was 45 and his children were, 22 and 16 years. They were all symptom-free. The reason for visiting genetic counseling was to ensure the health of his children. Also to find out the probable risk of APKD in the children's future, considering himself having been symptom free at their age. Moreover, he was concerned about the special care and considerations required to be applied in their life style, daily diet or the upcoming marriage-related issues.

It is worth noting that the 16-year-old daughter already had a suitor and they wanted to know whether they needed to discuss the case with him or that they could be assured of no risk for APKD in her future life. Furthermore, they were a highly religious family and visited the clinic just before Ramadan, the fasting month. Regarding the great interest of his children for fasting during this month, he wanted to know whether they were advised on the contrary.

Discussion

As PKD is an autosomal dominant disorder therefore each of the children have a 50% chance of being the carrier of the disease alleles. As mentioned before for diagnosing APKD imaging and genetic testing are the options of choice.

In testing of at-risk asymptomatic adults for ADPKD, the first step is renal image analysis (5). Molecular genetic testing is the other alternative. Because generalizations can be made about the phenotype expected in individuals with mutations in PKD1 versus PKD2, knowledge of the involved gene and causative mutation may provide valuable information on the level of disease severity in asymptomatic subjects (10).

Testing in the absence of definite symptoms of the disease is predictive testing. Renal imaging is regarded as the first step to test for ADPKD. Molecular genetic testing should be considered if the imaging results are equivocal or if a person aged <30yrs is looking for a definite diagnosis, as for a potential renal transplant donor (11). On the other hand, asymptomatic adult family members whom are at-risk may seek testing for making personal decisions for their future. Others may have different motivations such as the simple "need to know." Testing of these adult family members most often requires pre-test interviews to assess the motives for requesting the test, his/her knowledge of ADPKD, and the probable effect of positive and negative test results. All individuals seeking testing should be initially counseled regarding the possible issues which they may encounter including health, life, disability insurance coverage, employment prejudice, and changes in social and family interactions. For instance, individuals diagnosed with ADPKD should avoid playing certain sports such as football, since it may injure their kidneys. Other issues to consider are implications for the at-risk status of other family members. An informed consent should be signed and patient records kept confidential. Those with a positive test result will require further arrangements for long-term assessments and follow up.

In the present case, a genetic test could not be performed for the family's girl whom was 16 years old. Based on ethical rules and principles, genetic counseling in individuals under 18 years of age who are at-risk of non-curable adult-onset disease is not allowed and is considered unethical (12). The justification for this law is that performing the test before the age of 18 - the age when a person takes the authority of his own living in his own hands- could result in undesired or wrong decision-making by the family which is surely unethical.

Furthermore, there are certain concerns regarding the potential unhealthy adverse effects that such information may have on family dynamics, the risk of discrimination and stigmatization in the future, and the inevitable anxiety followed by receiving such information.

Experts in this field believe that in very young individuals, as the daughter of the current family, clinical monitoring should be performed as an alternative to genetic testing.

On the other hand, as the family had strong religious beliefs and were concerned about fasting in Ramadan, it should be mentioned that recent researches have indicated a negative impact for fasting in chronic kidney disease cases (13). It may also lead to major adverse cardiovascular events (MACE) (14). Therefore regarding all the aforementioned issues and the fact that the risk of PKD is high in similar individuals, fasting is not advised for such patients.

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