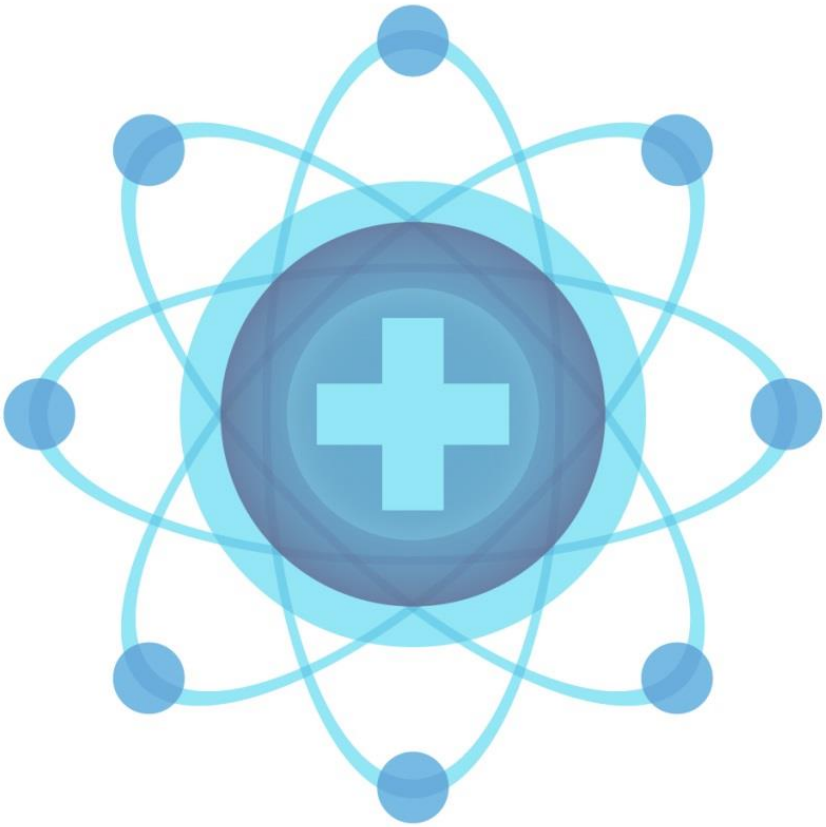

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GENEALOGICAL CHARACTERISTICS OF PATIENTS WITH CKD AT THE LEVEL OF PRIMARY HEALTH CARE

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Abstract: The main task of the genealogical method is to study the nature of the distribution of hereditary traits in the family. Genealogical analysis of pedigrees serves as the basis for medical and genetic counseling, that is, resolving the issue of the risk of having patients in a family burdened with hereditary diseases.

Keywords: Genealogical analysis, Transplantation, CKD, patients

Introduction. The most important method of preventing severe genetically determined diseases is medical and genetic counseling. Currently, clinical genetics uses various methods of obtaining information to identify patterns inherent in hereditary diseases. In clinical therapy, genealogical, mathematical-statistical and other methods of genetic analysis are often used. The main task of the genealogical method is to study the nature of the distribution of hereditary traits in the family. Genealogical analysis of pedigrees serves as the basis for medical and genetic counseling, that is, resolving the issue of the risk of having patients in a family burdened with hereditary diseases. When compiling pedigrees, special symbols are used. First of all, the so-called "legend" is drawn up, the initial collection of the family history is carried out. After drawing up the pedigree, a genealogical analysis is carried out.

Early diagnosis of chronic kidney disease (CKD) can help to reduce the progression of the disease, improve the clinical condition of patients and improve the prognosis of the course of the disease, reduce the number of patients with end-stage disease, reduce disability rates and reduce mortality. Based on the foregoing, the goal

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of our study was to study the role of genealogical analysis in patients with CKD as a method of prevention and early diagnosis of progression at the level of primary health care.

Materials and research methods. There were 217 CKD patients under observation in the primary health care unit, the average age of which was 46.17 ± 0.63 years, mainly of Uzbek nationality. The control group consisted of 20 healthy people of Uzbek nationality of the same age without signs of CKD.

After drawing up the legend of 217 families, a pedigree was compiled and a genealogical analysis was carried out. In order to identify the burden of CKD, the data obtained were compared with the results of the analysis of the pedigrees of individuals (n = 20) of a healthy population. The genealogical method was used to examine 893 relatives of the 1st degree of relationship (parents, siblings) of patients with CKD for the presence of renal pathology. A detailed pedigree was compiled, which included information about diseases in 2-3 generations of the family. Genetic material was collected from both parental lines by cross-examination of both parents, sometimes grandparents. A total of 1761 people were analyzed in the model population.

Results and its discussion. The data obtained were compared with the generalized family response of 20 practically healthy people, in the model population of which 172 people were analyzed, of which the incidence of renal pathology was 2.13%. Comparative analysis of the generalized family portrait of CKD patients with a generalized family portrait of a population of practically healthy people revealed a more frequent lesion of renal pathology in relatives with CKD (11.07 %). In families of probands, renal pathology in generations in relation to the total number of patients with each concentration is III - 7.49%; II - 11.06%; I - 17.43%. The burden in the population of practically healthy people was observed significantly less: III - 0%; II - 0.15%; I- 0%.

It turned out that relatives of the 1st degree of kinship 130 (14.56 ± 2.50%) suffer from renal pathology more often, which in relation to the total number of

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patients with this concentration is 7.38%. At the same time, the following distribution was observed among groups: in group 1 (n = 224) - in 27 people, which amounted to 12.05 ± 0.69%, of which 16 men (59.26 ± 0.57%), 11 women (40.74 ± 0.41%); Group 2 (n = 219) - 28 (12.78 ± 0.75%), of which 19 men (67.86 ± 0.68%), 9 women (32.14 ± 0.38%); Group 3 (n = 239) - 36 (15.06 ± 0.81%), of which 23 men (63.89 ± 0.62%), 13 women (36.11 ± 0.50%); Group 4 (n = 211) - 39 (18.48 ± 0.76%), of which 22 men (54.41 ± 0.64%), 17 women (43.59 ± 0.51%). Hereditary burden of renal pathology was observed more often on the father's side in 46 (10.60 ± 1.50%) than on the mother's side 19 (4.38 ± 1.04%) (Table 1). Among the 1st degree relatives of grandmothers (n = 434) with renal pathology, there were 29 (6.68 ± 1.22%) of them on the father's side 18 (62.07±0.93%), on the mother's side 11 (37,93 ± 0.73%).

Table 1.

The incidence of CKD in the pedigrees of CKD patients.

Study group		Prevalence of renal disease in relatives of patients
All relatives n = 1761	Total	195(11,07±3,14%)
	Men	116(59,48±2,46%)
	Women	79(40,51±1,91%)
Mothers n = 217		20(9,22±0,98%)
Fathers n = 217		28(12,90±1.13%)
Grannies n = 434	Total	29(6,68±1,22%)
	From the mother's side	11(37,93±0,73%)
	From the father's side	18(62,07±0,94%)
Grandfathers n = 434	Total	36(8,29±1,29%)
	From the mother's side	8(22,22±0,62%)
	From the father's side	29(77,78±1,13%)
Sibs n = 459	Total	80(17,43±1,98%)
	Men	51(63,75±1,67)
	Women	29(36,25±1,15)
Relatives of the 1st degree of kinship n = 893	Total	130(14,56±2,50%)
	Men	80(61,54±2,08%)
	Women	50(38,46±1,49%)

Let us consider their distribution into groups: in group 1 (n = 54) - in 2 people, which amounted to 1.85 ± 0.19%, of them on the father's side 1 (50 ± 0.14%), on the

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mother's side - 1 (50 ± 0.14%); Group 2 (n = 105) - 5 (4.71 ± 0.30%), of which on the father's side 3 (60 ± 0.23%), on the mother's side - 2 (40 ± 0.19%); Group 3 (n = 116) - 8 (6.90 ± 0.35%), of which 6 on the father's side (75 ± 0.31%), on the mother's side - 2 (25 ± 0.18%); Group 4 (n = 104) - 14 (13.46 ± 0.53%), of which 8 on the father's side (57.14 ± 0.36%), on the mother's side - 6 (42.86 ± 0.32 %). There were 36 grandfathers (n = 434) with renal pathology (8.29 ± 1.29%), 28 of them on the father's side (77.78 ± 1.13%), on the mother's side 8 (22.22 ± 0.62 %). Let us consider their distribution into groups: in group 1 (n = 54) - in 4 people, which amounted to 3.70 ± 0.26%, of which all were not observed on the part of the father on the part of the mother; Group 2 (n = 105) - 7 (6.60 ± 0.34%), of which 6 on the father's side (85.71 ± 0.32%), on the mother's side - 1 (14.29 ± 0.14 %); Group 3 (n = 116) - 10 (8.62 ± 0.38%), of which 8 on the father's side (80 ± 0.35%), on the mother's side - 2 (20 ± 0.18%); Group 4 (n = 104) - 15 (14.42 ± 0.50%), of which 10 on the father's side (66.67 ± 0.40%), on the mother's side - 5 (33.33 ± 0.30 %). In a genealogical study, 459 siblings were studied, of which renal pathology was noted in 80, which amounted to 17.49 ± 1.98%. At the same time, 51 of them were men (63.75 ± 1.67%), and 29 women (36.25 ± 1.15%). When examined by groups, the following picture was observed: in group 1 (n = 116) - in 19 people, which amounted to 16.38 ± 0.59%, of which 11 men (57.89 ± 0.53%), women - 8 (42.11 ± 0.36%); Group 2 (n = 113) - 20 (17.70 ± 0.66%), of which 14 men (70 ± 0.56%), 6 women (30 ± 0.32%); Group 3 (n = 123) - 20 (16.26 ± 0.58%), of which 14 men (70 ± 0.47%), 6 women (30 ± 0.31%); Group 4, (n = 107) - 21 (19.63 ± 0.60%), including 12 men (57.15 ± 0.51%), women - 9 (42.85 ± 0.43%).

For illustration, we give an example: *pedigree of the patient L.F., 53 years old; Diagnosis: basic Ischemic heart disease voltage angina, FC1. GD 2, AG 2, H 3; Competed - chronic glomerulonephritis, mixed form, diabet mellitinus 2 type, severe current, in the decompensation stage; Complication - CKD 3Ast. Diabetic encephalopathy, diabetic nephropathy.* (Fig.1) From the pedigree it can be seen that the patient has a hereditary burden on the part of the patient, the patient has a

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hereditary burden on the part of the father, whose grandfather suffered from renal pathology, on the maternal side - no morbidity was detected.

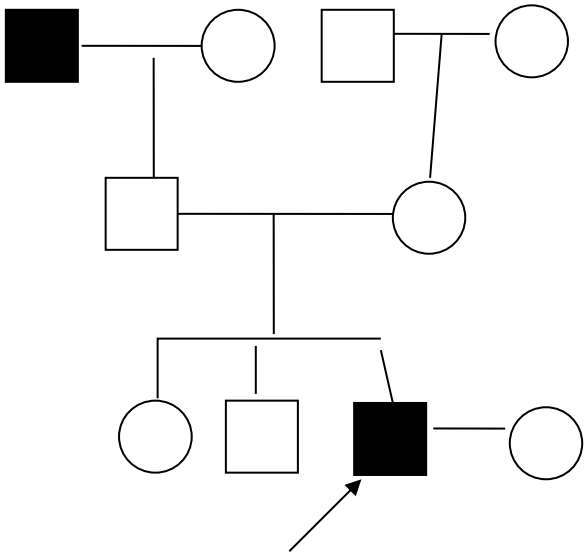

Fig. 1. Fragment of L.F.'s pedigree, 53 years old.
Note: ■ - a patient with CKD.

The more frequent lesion of CKD in the relatives of the proband than in the relatives of healthy people indicates a significant role of genetic factors in its occurrence.

The results of our study indicate a significant prevalence of CKD in families of probands, but with a predominant lesion of relatives of the first degree of relationship. The analysis of the pedigree confirms the absence of a monogenously inherited disease as a simple recessive or dominant inheritance. Along with this, the wide spread of renal pathology in the population suggests that only a predisposition to the development of CKD is inherited. A decrease in the incidence of CKD in relatives with a distance from the degree of relationship from the proband may indicate the multifactorial nature of the disease. Such diseases are called multifactorial or polygenic, since their occurrence is due to the action of a whole complex of genes and the influence of the external environment.

Output. The results of the study showed that CKD can be considered as having a hereditary predisposition, which brings it closer to polygenic diseases. Taking this

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into account, the use of methods of clinical and genetic examination of patients in primary health care should be more widely recommended, since knowledge of the family background greatly facilitates the early diagnosis and prevention of disease progression. Moreover, the hereditary burden was more on the paternal side. At the same time, nephropathy was more often registered in the relatives of the proband of the I-II degree of kinship in comparison with other relatives. A decrease in the frequency of renal pathology in relatives with a distance from the degree of relationship from the proband may indicate the multifactorial nature of the disease. The risk of developing the disease was higher in cases when the father or mother of the proband suffered from nephropathy.

Thus, CKD can be considered as having a hereditary predisposition, which brings it closer to polygenic diseases. The polygenicity hypothesis explains well why in families with a large number of affected members, CKD is difficult, becomes protracted, and often progresses to the terminal stage. Taking this into account, the use of methods of clinical and genetic examination of patients should be more widely recommended, since knowledge of the family background greatly facilitates the prognosis of individual cases of the disease within the family.

Moreover, it is more accessible precisely in the primary health care setting from family doctors.

10.5281/zenodo.5547926

References:

1. Abdulloev S.M., Gulov M.K. Clinical and epidemiological features and risk factors for the development of chronic kidney disease in the Republic of Tajikistan // Healthcare of Tajikistan. - 2019. - No. 2. - S. 5-13.

2. Akhmedova N.Sh. Features of screening of renal function in an outpatient setting // Medicus. - 2019. - No. 2 (26). - S. 17-21.

3. Veltischev Yu.E., Ignatova M.S. Preventive and preventive nephrology (genetic and ecopathogenic risk factors for the development of nephropathy): A guide for doctors. - M., 1996. - with. 61

4. Ignatova M.S. The role of genetic research in the development of nephrology // Therapeutic archives. - M., 2003. - No. 6. - P.66-72.

5. Podzolkov V.I., Bragina A.E. Chronic kidney disease as a multidisciplinary problem of modern medicine // Therapeutic archive. - 2018. - T. 90, No. 6. - P.121-129.

6. Bisi-Onyemaechi AI, Okafor HU, Ughasoro MD. Profile of chronic kidney disease modifiable risk factors in a rural community of south east Nigeria. // BMC Public Health. 2018 Jul 27; 18 (1): 922.

7. Connaughton DM, Kennedy C, Shril S, Mann N, Murray SL, Williams PA, Conlon E, Nakayama M, van der Ven AT, Ityel H, Kause F, Kolvenbach CM, Dai R, Vivante A, Braun DA, Schneider R, Kitzler TM, Moloney B, Moran CP, Smyth JS, Kennedy A, Benson K, Stapleton C, Denton M, Magee C, O'Seaghdha CM, Plant WD, Griffin MD, Awan A, Sweeney C, Mane SM, Lifton RP, Griffin B, Leavey S, Casserly L, de Freitas DG, Holian J, Dorman A, Doyle B, Lavin PJ, Little MA, Conlon PJ, Hildebrandt F. Monogenic causes of chronic kidney disease in adults. Kidney Int. 2019 Apr; 95 (4): 914-928. doi: 10.1016 / j.kint.2018.10.031. Epub 2019 Feb 14. PMID: 30773290; PMCID: PMC6431580.

8. Gaitonde DY, Cook DL, Rivera IM. Chronic Kidney Disease: Detection and Evaluation. // Am Fam Physician. 2017 Dec 15; 96 (12): 776-783.

10.5281/zenodo.5547926
9. Groopman EE, Povysil G, Goldstein DB, Gharavi AG. Rare genetic causes of complex kidney and urological diseases. Nat Rev Nephrol. 2020 Nov; 16 (11): 641-656. doi: 10.1038 / s41581-020-0325-2. Epub 2020 Aug 17. PMID: 32807983; PMCID: PMC7772719.

10. Hwang, Ji-Yun et al. "Family history of chronic renal failure is associated with malnutrition in Korean hemodialysis patients." Nutrition research and practice vol. 3.3 (2009): 247-52. doi: 10.4162 / nrp.2009.3.3.247

11. McClellan WM, Satko SG, Gladstone E, Krisher JO, Narva AS, Freedman BI. Individuals with a family history of ESRD are a high-risk population for CKD: implications for targeted surveillance and intervention activities. Am J Kidney Dis. 2009 Mar; 53 (3 Suppl 3): S100-6. doi: 10.1053 / j.ajkd.2008.07.059. PMID: 19231753.

12. Malik S, Syed Z, Naz F, Rehman N, Rauf A, Ali R. Risk Factors Of Chronic Kidney Disease Leading To Dialysis In Patients Presenting At Ayub Teaching Hospital Abbottabad. // J Ayub Med Coll Abbottabad. 2019 Oct-Dec; 31 (Suppl 1) (4): p 672-673.