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The identification of single nucleotide polymorphisms (SNPs) in patients who have chronic

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ABSTRACT

Background Occurring at a frequency of about one every 300 base pairs, SNPs are the most abundant form of genetic variation in the human genome. Every single one of us possesses a one-of-a-kind fingerprint made up of one of over 100 million SNPs. **Methodology** This study was carried out on 200 patients with chronic diseases that fulfilled the sampling criteria according to the research objective. Many chronic diseases included heart, blood pressure, diabetes, arthritis, and other diseases that were diagnosed clinically and through many tests. In contrast, there was another group of unaffected control people. Many tests were conducted for them, and after confirming these diseases, blood samples were drawn for genetic tests. **Results** the number of single nucleotide polymorphisms and their relationship to chronic diseases. The frequency of the genetic mutation (rs7903146) associated with the TCF7L2 gene was observed at a rate of 31 at the AA base and a risk ratio of 1.62 in people with diabetes, while the frequency of the mutation (rs10757274) associated with the CDKN2 gene was observed at a rate of 37 at the nitrogenous base. GG with a risk rate of 2.2 in people with heart disease. Finally, the rs2476601 mutation associated with the PTPN22 gene was repeated at a rate of 34% in TT with a risk rate of 2.1.

Keywords: Single nucleotide polymorphisms (SNPs) , chronic diseases

INTRODUCTION

One kind of genetic variation that can happen within DNA sequences is known as single nucleotide polymorphism (SNP) [1]. When one nucleotide is substituted for another, for example, when A is replaced with C, G, or T, this is called a single nucleotide polymorphism [2]. Distinctions in naturally occurring DNA sequences between people provide a potent window into the genetic underpinnings of human health and illness [3]. Changes in just one base pair of DNAS, called single nucleotide polymorphisms (SNPs), can influence gene function and environmental interactions [4]. At about one occurrence every 300 base pairs, single-nucleotide polymorphisms (SNPs) constitute the vast majority of genetic variation in the human genome [5]. Every single one of us possesses a one-of-a-kind fingerprint made up of one of over 100 million SNPs [6]. Because SNPs affect sensitivity, severity, progression, and response to treatment for a wide range of diseases and disorders, they can have significant consequences for comprehending disease risk [7]. Diseases like cystic fibrosis and sickle cell anemia, which arise from mutations in a single gene, can be directly caused or prevented by some SNPs [8]. Complex disorders like cancer, diabetes, and Alzheimer's can have their causes traced back to other SNPs that alter gene expression, protein activity, or the way these components interact with one

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another. [9] The rate of caffeine metabolism, its effects on blood pressure and heart rate, and other parameters can vary from one individual to the next due, for instance, to variations in the gene encoding the enzyme responsible for these processes.

METHODOLOGY

STUDY POPULATION

This research was done on 200 patients who had chronic diseases and fulfilled the criteria of sampling set by the purpose of the research. Many chronic diseases included heart disease, high blood pressure, diabetes, arthritis, and other diseases that were diagnosed clinically and through many tests. In contrast, there was another group of unaffected control people. Many tests were conducted for them, and after confirming these diseases, blood samples were drawn for genetic tests.

DETECTION OF SNP BY PCR

Polymorphisms in a sample are identified by amplification of isolated genomic DNA by polymerase chain reaction (PCR) and fragmentation to enhance probe binding efficiency. The DNA is subsequently labelled with a fluorescent marker, and it is subsequently hybridized to the slide [10, 11].

RESEARCH CONTENT

The general information characteristics of the research subjects are recorded: age, sex, height, weight, BMI, education level, pulse, systolic, and diastolic blood pressure in this study. Single nucleotide polymorphisms

DATA ANALYSIS

The results were analyzed using SPSS and SAS [12].

RESULTS AND DISCUSSION

Table 1. Relationships between age and chronic diseases

		Age			
		20-40	40-50	50-70	Total
Disease	Diabetic	<5	10	35	49
	Heart Disease	<5	15	30	48
	Kidney Disease	<5	15	35	53
	Bone Disease	5	10	35	50
Total		15	50	135	200

Table 1 illustrates that specific age categories are linked to specific chronic diseases. The age groups of 40–50 and 50–70 are disproportionately affected by chronic diseases, including diabetes, heart disease, and osteoarthritis, with the latter two suffering the highest rates of occurrence. Conversely, the age group of 20–40 exhibited the lowest rates of occurrence. The natural aging process results in the accumulation of various substances in the interior of blood vessels. As a result, it is less elastic and

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malleable than it was during its youth. The accumulation of these compounds, which results in atherosclerosis, increases the likelihood of heart attacks and other vascular complications [13].

Table 2. Chi-square of relationships between age and chronic diseases

Chi-Square Tests			
	Value	df	Asymptotic Significance (2-sided)
Pearson Chi-Square	3.032[a]	6	.805
Likelihood Ratio	3.021	6	.806
Linear-by-Linear Association	.020	1	.887
N of Valid Cases	200		

a. 4 cells (33.3%) have an expected count less than 5. The minimum expected count is 3.60.

The chi-square value for the relationship between age group and chronic diseases is presented in Table 2. There were no discernible distinctions between the age categories and the types of maladies. This outcome can be attributed to the convergence of the prevalence of chronic diseases among various age groups, as illustrated in Figure 1

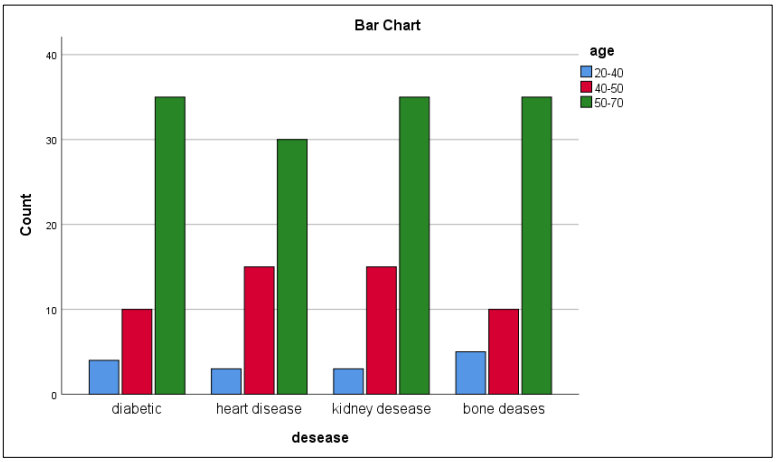

Fig. 1. Relationships between age and chronic diseases

Table 3. Relationships between gender and chronic diseases

Count

		Gender		
		Male	Female	Total
Disease	Diabetic	24	23	47
	Heart Disease	27	22	49
	Kidney Disease	11	38	49
	Bone Disease	12	43	55
Total		74	126	200

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Gender is associated with hereditary disorders (Table 3). It was shown that there's a wide variation in susceptibility between men and women for the various diseases. Men are more likely to suffer from diabetes and heart disease, while women are more likely to suffer from arthritis and bone infections. Not only that, but they're more prone. In cases of kidney and urinary tract infections in particular. Because of inherent differences in their biology, women have a disproportionate number of health problems compared to men. Headaches and osteoporosis are the two most prevalent of these conditions.

Diseases that impact both sexes are only one more way in which men and women differ from one another. For example, compared to men, women get migraines three times more frequently 18% vs. 6%) and headaches in general [13].

Table 4. Chi-square of relationships between gender and chronic diseases

Chi-Square Tests

	Value	df	Asymptotic Significance (2-sided)
Pearson Chi-Square	20.766[a]	3	0.000
Likelihood Ratio	21.137	3	0.000
Linear-by-Linear Association	15.616	1	0.000
N of Valid Cases	200		

a. 0 cells (0.0%) have an expected count less than 5. The minimum expected count is 17.39.

Through the results obtained in Table 4, the Chi-square values for the relationship between gender and chronic diseases were found. There were significant differences between all diseases and gender, as the value of Chi-square was 20.7. Thus, there were significant differences for all diseases with gender, as shown in Figure 2. This situation can be explained by the reason for the biological composition of women's bodies: it is that they are more susceptible to back pain than men, and figures indicate that women are two-thirds more likely to suffer from osteoarthritis than men. Symptoms may differ between the sexes. The symptoms of a heart attack are different in women than in men, and the most common symptoms in women are nausea and back pain, so doctors usually delay in diagnosing heart attacks in women [14, 15].

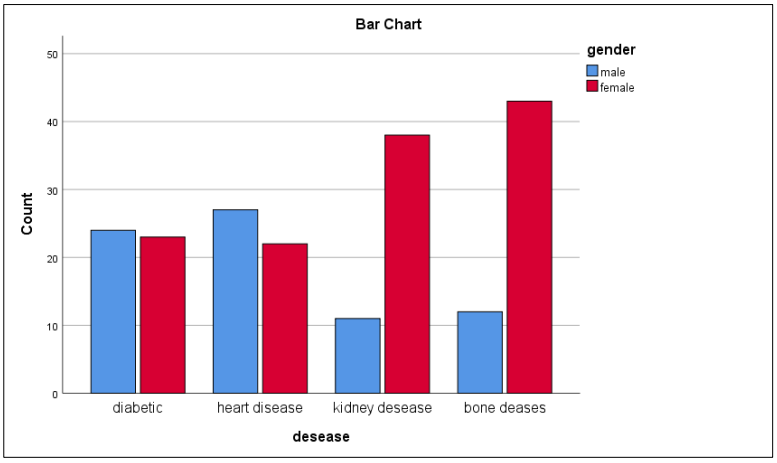

Fig. 2. Relationships between gender and chronic diseases

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Table 5. The percentage of single nucleotide polymorphisms for some chronic diseases

Chronic Disease	SNP	Associated Gene	Percentage	OR	P-value
Diabetic	**rs7903146**	TCF7L2	31 AA	1.62	0.003
Heart disease	**rs10757274**	CDKN2	37 GG	2.2	0.004
Rheumatoid arthritis	**rs2476601**	PTPN22	34 TT	2.1	0.001

Table 5 shows the number of single nucleotide polymorphisms and their relationship to chronic diseases. The frequency of the genetic mutation (rs7903146) associated with the TCF7L2 gene was observed at a rate of 31 at the AA base and a risk ratio of 1.62 in people with diabetes, while the frequency of the mutation (rs10757274) associated with the CDKN2 gene was observed at a rate of 37 at the nitrogenous base. GG with a risk rate of 2.2 in people with heart disease. Finally, the rs2476601 mutation associated with the PTPN22 gene was repeated at a rate of 34% in TT with a risk rate of 2.1. We conclude from these results that chronic diseases have the greatest number of mutations, which vary in severity depending on these diseases, as heart diseases took the lead in This poses a risk compared to other diseases. These mutations may be explained by the type and quantity of medications taken by patients. As in Figure 3.

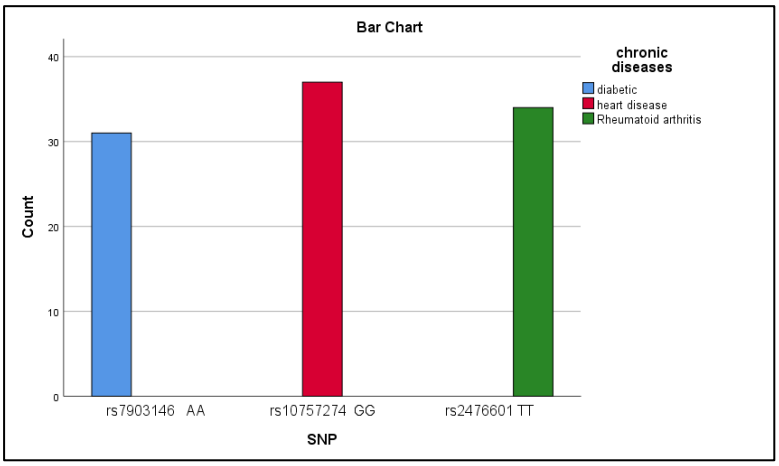

Fig. 3. Percentage of single nucleotide polymorphisms for some chronic diseases

CONCLUSION

The study indicates the significance of SNPs in the development and progress of a number of chronic diseases. In the study, critical genetic mutations were traced in the analysis of 200 patients with chronic conditions like diabetes, heart disease, arthritis, and many other disorders against controls. The results have highlighted that, for instance, rs7903146 (TCF7L2 gene), rs10757274 (CDKN2 gene), and rs2476601 (PTPN22 gene) are strongly associated with increased risks for diabetes, heart disease, and other chronic diseases, respectively. Such findings support the view that genetic predispositions stand in an important position regarding disease manifestation and severity and, among the diseases under study, heart diseases have the highest genetic risk.

The study also established age and gender as important variables in the prevalence and distribution of chronic diseases. Individuals aged 40–70 years were most affected by chronic conditions, presumably due to physiological changes accompanying aging. There were also clear gender disparities, with men being more susceptible to diabetes and heart disease, while women were more prone to arthritis, migraines, and urinary infections, mainly because of biological and hormonal factors. The findings also reiterated that women have different symptoms for heart attacks and other such ailments, which requires a different method of diagnosis.

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REFERENCES

[1] J.-L. C. Mougeot *et al.*, "Identification of single nucleotide polymorphisms (SNPs) associated with chronic graft-versus-host disease in patients undergoing allogeneic hematopoietic cell transplantation," *Supportive Care in Cancer*, vol. 31, no. 10, Sep. 2023, doi: 10.1007/s00520-023-08044-3.

[2] Yu. E. Azarova *et al.*, "Polymorphisms of the NCF4 Gene Increase the Risk of Chronic Heart Failure in Patients with Type 2 Diabetes Mellitus," *Bulletin of Experimental Biology and Medicine*, vol. 176, no. 1, pp. 77–81, Nov. 2023, doi: 10.1007/s10517-023-05974-0.

[3] A. Del Cuore *et al.*, "Metabolic memory in diabetic foot syndrome (DFS): MICRO-RNAS, single nucleotide polymorphisms (SNPs) frequency and their relationship with indices of endothelial function and adipo-inflammatory dysfunction," *Cardiovascular Diabetology*, vol. 22, no. 1, Jun. 2023, doi: 10.1186/s12933-023-01880-x.

[4] S. Perumalsamy, H. Z. Huri, B. M. Abdullah, O. Mazlan, W. A. W. Ahmad, and S. R. D. B. Vethakkan, "Genetic Markers of Insulin Resistance and Atherosclerosis in Type 2 Diabetes Mellitus Patients with Coronary Artery Disease," *Metabolites*, vol. 13, no. 3, p. 427, Mar. 2023, doi: 10.3390/metabo13030427.

[5] N. M. Kulaiets, "Association of β1- and β2-adrenergic receptor gene polymorphisms with the risk of heart failure in patients with type 2 diabetes mellitus," *INTERNATIONAL JOURNAL OF ENDOCRINOLOGY (Ukraine)*, vol. 20, no. 4, pp. 297–306, Aug. 2024, doi: 10.22141/2224-0721.20.4.2024.1409.

[6] S. Ashiq and M. F. Sabar, "Association of maternal hypertension and diabetes with variants of the NKX2-5, LEFTY1 and LEFTY2 genes in children with congenital heart defects: a case-control study from Pakistani Population," *Molecular Biology Reports*, vol. 50, no. 6, pp. 5013–5020, Apr. 2023, doi: 10.1007/s11033-023-08418-0.

[7] X. Li *et al.*, "Shared Genetic Architectures between Coronary Artery Disease and Type 2 Diabetes Mellitus in East Asian and European Populations," *Biomedicines*, vol. 12, no. 6, p. 1243, Jun. 2024, doi: 10.3390/biomedicines12061243.

[8] W.-W. Chang *et al.*, "Association of tag single nucleotide polymorphisms (SNPs) at lncRNA MALAT1 with type 2 diabetes mellitus susceptibility in the Chinese Han population: A case-control study," *Gene*, vol. 851, p. 147008, Oct. 2022, doi: 10.1016/j.gene.2022.147008.

[9] T.-W. Wu, C.-L. Chou, C.-F. Cheng, S.-X. Lu, Y.-J. Wu, and L.-Y. Wang, "Associations of genetic markers of diabetes mellitus with carotid atherosclerosis: a community-based case–control study," *Cardiovascular Diabetology*, vol. 22, no. 1, Mar. 2023, doi: 10.1186/s12933-023-01787-7.

[10] C. A. Hageh, S. O'Sullivan, S. Chacar, A. Henschel, M. Nader, and P. Zalloua, "CORONARY ARTERY DISEASE PATIENTS WITH RS7904519 (TCF7L2) ARE AT a PERSISTENT RISK OF TYPE 2 DIABETES," *Journal of the American College of Cardiology*, vol. 83, no. 13, p. 4594, Apr. 2024, doi: 10.1016/s0735-1097(24)06584-7.

[11] M. Merzah, S. Natae, J. Sándor, and S. Fiatal, "Single Nucleotide Variants (SNVs) of the Mesocorticolimbic System Associated with Cardiovascular Diseases and Type 2 Diabetes: A Systematic Review," *Genes*, vol. 15, no. 1, p. 109, Jan. 2024, doi: 10.3390/genes15010109.

[12] Y. Zhao *et al.*, "Type 1 diabetes, its complications, and non-ischemic cardiomyopathy: a mendelian randomization study of European ancestry," *Cardiovascular Diabetology*, vol. 23, no. 1, Jan. 2024, doi: 10.1186/s12933-023-02117-7.

[13] D. Petrovič, P. Nussdorfer, and D. Petrovič, "The rs3825807 Polymorphism of ADAMTS7 as a Potential Genetic Marker for Myocardial Infarction in Slovenian Subjects with Type 2 Diabetes Mellitus," *Genes*, vol. 14, no. 2, p. 508, Feb. 2023, doi: 10.3390/genes14020508.

[14] A. Mansour, M. Mousa, D. Abdelmannan, G. Tay, A. Hassoun, and H. Alsafar, "Microvascular and macrovascular complications of type 2 diabetes mellitus: Exome wide association analyses," *Frontiers in Endocrinology*, vol. 14, Mar. 2023, doi: 10.3389/fendo.2023.1143067.

[15] A. Del Cuore *et al.*, "Metabolic Memory in Diabetic Foot Syndrome (DFS): Epigenetic Changes of the Expression of Micro-rnas and Single Nucleotide Polymorphisms (SNPS) Frequency in a Cohort of Diabetic Patients With and Without Foot Ulceration and Correlation With Indices of Endothelial and Adipo-inflammatory Dysfunction," *Research Square (Research Square)*, Apr. 2023, doi: 10.21203/rs.3.rs-2814460/v1.

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