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RESEARCH ARTICLE

STUDY ON AWARENESS ABOUT GENETIC DISEASES AMONG YOUTH POPULATION IN AKOLA CITY OF MAHARASHTRA

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ABSTRACT

Background: Genetic diseases forms sizeable portion of diseases which being an important cause of morbidity and mortality, they impose a heavy burden on families and the health sector in our country. Thus Awareness about genetic diseases is crucial in today's world. Aim and Objectives: To study awareness about genetic diseases among youth population in Akola city and to gain an insight into their understanding about it. Methodology: The cross-sectional study was conducted among 130 youths of Akola city in the month of March and April 2022. A predesigned pretested schedule was used to collect information on socio-demographic profile, knowledge, attitude and practices about genetic diseases was observed through interviews. Results: Majority of participants were male, 88.46% were in the age group of 20-30 years, 80% were studied graduation and above, 61.54% were students, 82.30% participants were aware about genetic counselling, 67.69% disagree that late marriages increases the risk of genetic diseases, 85.38% participants were interested to screen both themselves and their partner before marriage, 71.53% participants inquired about presence of genetic diseases in the family of person they wish to marry and 63.85% participants check blood group of partner before marriage, 91.53% participants accepted consanguineous marriage as a cause of genetic disease. Conclusion: Although majority of participants were educated and aware of genetic diseases, very few translated it in their daily practices. Pre-marital counselling and Screening should be made mandatory which in turn can reduce genetic diseases. Practical Application: This study can help to train and formulate policies and create awareness about Genetic diseases and can help them make better choices in their lives.

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INTRODUCTION

According to National Human Genome Research Institute, A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorderhaemoglobinopathies, cystic fibrosis and haemophilia) and chromosomal disorders (Down syndrome, among others) by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes

(changes in the number or structure of entire chromosomes, the structures that carry genes). Some diseases are caused by mutations that are inherited from the parents and are present in an individual at birth, like sickle cell disease. Other diseases are caused by acquired mutations in a gene or group of genes that occur during a person's life. Such mutations are not inherited from a parent, but occur either randomly or due to some environmental exposure (such as cigarette smoke). Some genetic diseases, such as haemophilia, are carried on the X-chromosome (these X-linked disorders occur mainly in men).

Others can arise from the presence of an abnormal gene in any autosome: if the gene is dominant, it results always in what is called a dominant condition, whereas if it is recessive many of these diseases appear only when the gene is inherited from both parents (and are thus called recessive conditions)².

AIM & OBJECTIVES

The main objective of this study is to investigate and understand awareness about genetic diseases among youth population in Akola city in Maharashtra state.

MATERIAL AND METHODS

Study design and duration: This was a cross-sectional study conducted in Akola city with a view to assessing the level of awareness about genetic diseases among youth population

Study setting: The study was conducted in the densely populated areas of Akola city of Maharashtra state in India. (Wikipedia/Akola district map.)



Sample size estimation: Sample size taken as 130.

Study Period: The survey was conducted in the month of March and April 2022.

Sampling technique: Non-probability convenient sampling

Study population: Youth population of the of Akola city

Inclusion criteria: Inclusion criteria were those who agreed to Participate in the study.

Exclusion criteria: Those who were unwilling to participate and person with age less than 20 years and more than 40 years of age were excluded from our study.

Study tool: Predesigned, pretested schedule was used.

Instrument development and data collection procedure: An English version of the questionnaire was developed. The questionnaire was divided into two parts consisting of sociodemographic profile, knowledge, attitude, practice about genetic diseases. ^{3,4,5,11,13,14,15,16}

Part I is designed to determine "socio-demographic profile including age, gender, education, blood group, drinking and smoking habits, residence and income.

Part II comprises of 20 Questions to assess knowledge, attitudes and practices.

RESULTS AND DISCUSSION

Socio-demographic profile of participants: In Table-1 are shown socio-demographic profile of participants, which shows that most (65.38%) of the 130 respondents were male similar study conducted among students of Saudi Arabia show contrasting results showing high females.¹⁶ and 88.46% (n=115) participants age between 20 and 30 years similar findings were reported in Saudi arabia¹⁷. About 61.54% participants were students and 72.30% had no income. 80% of respondents completed their higher secondary education and studied till post-graduation while 19.99% were in below higher secondary similar findings were reported in Saudi arabia. 5,17 46.92% of participants had B+ve blood group while 18.46% had O+ve blood group. Around 13.84% and 10.74% of participants practiced drinking, smoking and tobacco chewing respectively. Out of 130 participants 65.38% and 34.61% resided in Rural and Urban areas respectively. Out of 130 participants only 0.77% (n=1) suffered from genetic disease.

Participant's knowledge towards genetic diseases: Majority (94.61%) of participants were aware that genetic disease were inherited from biological father and mother while 91.54% of participants agreed that consanguineous marriage is leading cause of genetic disease similar findings were reported in Saudi Arabia. 17 while 4.62% considered inter-caste marriage as a cause. 81.53% participants identified Albinism as genetic disorder while 6.15% considered vitamin deficiency as genetic disease. 70.77% of participants considered all the three unnecessary exposure to X-rays, consumption of LSD drug and excessive alcohol consumption during pregnancy as a cause of genetic abnormality. 28.46% and 28.4649.23% of participants considered Rh positive mother and Rh negative foetus and Rh negative mother and Rh positive foetus cause of Erythroblastosis fetalis respectively. 82.31% were aware that hemophilia is hereditary blood disorder while 8.46% considered it as disease affecting mental health. 89.23% were aware about Down syndrome which affects mental and physical health while 4.61% considered it as congenital disease of kidney. 83.07% and 81.53% of participants were aware about sickle cell anemia as condition with abnormal hemoglobin and thalassemia as blood disease respectively reported similar findings were in Odisha BasantakumarBindhani*et al*¹⁵. 24.61% of participants considered phenylketonuria as inherited kidney disease while 57.70% considered deficiency of liver enzyme. 54.61% considered cystic fibrosis as disease affecting respiratory and intestinal system.

Participant's Attitude and Practices towards genetic diseases: Out of 130 participants 67.69% disagreed with idea that late marriages increases risk of genetic diseases while 32.30% agreed with it. 85.38% agreed to screen themselves and their partner with whom they wish to marry for genetic disease before marriage similar findings were reported in Odisha by Basantakumar Bindhani et al¹⁵ as 7.69% were not of opinion for carrying out screening, 3.85% considered screening only for partner. 71.53% enquired about presence of genetic disease in family of person with whom they wish to marry while 28.46% did not enquire. 63.85% agreed to check blood group of their partner before marriage while 36.15% disagreed similar findings were reported in Saudi Arabia.¹⁷ 70% considered all the three reasons to screen one self and one's partner before marriage as it can reduce the frequency of genetic disease, it can reduce the cost of expenditure on

Table 1. Socio-Demographic Profile Of Participant

CHARACTERISTICS	CATEGORIES	FREQUENCY (n)=130	PERCENTAGE (%)
AGE (YEARS)	20-24	50	38.46
	25-30	65	50.00
	31-39	10	07.69
	40 AND ABOVE	05	03.84
GENDER	MALE	85	65.38
	FEMALE	45	34.61
EDUCATION	UPTO 10 th CLASS	03	02.30
	UPTO 12th CLASS	23	17.69
	UPTO GRADUATION	65	50.00
	UPTO POST GRADUATION	33	25.39
	ABOVE POST GRADUATION	06	04.61
OCCUPATION	STUDENT	80	61.54
	FARMER	03	02.31
	GOVERNMENT SERVANT	08	06.15
	ENGINEER	05	03.85
	DOCTOR	06	04.61
	ADVOCATE	06	04.61
	PRIVATE JOB	16	12.31
	BUSINESSMEN	06	04.61
BLOOD GROUP	A+	18	13.84
BLOOD GROUP	B+	61	46.92
	AB+	13	10.00
	А Б ⁺ О+	24	18.46
	A-	01	
	A- B-	01	00.77
			00.77
	AB-	01	00.77
	O-	01	00.77
Digo. E	NOT AWARE	10	07.69
INCOME	NO INCOME	94	72.30
(PER MONTH)	RS. 5000 TO 30,000	16	12.30
	RS. 31,000 TO 70,000	09	06.92
	RS. 71,000 TO 1,00,000	03	02.30
	RS. 1,00,001 AND ABOVE	08	06.15
RESIDENCE	RURAL	85	65.38
	URBAN	45	34.61
DRINKING	NO	112	86.15
	YES OCCASIONALLY	18	13.84
	YES HABITUAL	00	00.00
SMOKING/CHEWING O	F NO	116	89.23
TOBACCO/GUTKHA	YES OCCASIONALLY	09	06.92
	YES HABITUAL	05	03.84
ARE YOU SUFFERING FROM	M NO	129	99.23
ANY GENETICS DISEASE?	YES	01	00.77
	SICKLE CELL ANEMIA		
	THALASSEMIA		
	HAEMOPHILIA		
	DOWN SYNDROME		
	G6 PD DEFICINCY		

Table 2. Knowledge, Attitude and Practices About Genetic Diseases Among Youth Population

	OPTIONS	FREQUENCY	PERCENTAGE
		(n)=130	(%)
WHAT IS MEANT BY GENETIC	*DISEASES RELATED TO BONE	02	01.54
DISEASE ?	*DISEASES INHERITED FROM BIOLOGICAL FATHER AND	123	94.61
	MOTHER		
	*DISEASE OF BLOOD	05	03.85
	*DISEASE RELATED TO SKIN	00	00.00
WHAT DO YOU THINK IS CAUSE OF GENETIC DISEASE ?	*CONSANGUINOUS MARRIAGE	119	91.53
	*INTER CASTE MARRIAGE	06	04.62
	*INTER RELIGIOUS MARRIAGE	03	02.30
	*ACCIDENT	02	01.53
WHICH OF THE FOLLOWING DISEASE IS INHERITORY ?	*ALBINISM	106	81.53
	*VITAMIN DEFICIENCY	08	06.15
	*ANEMIA	14	10.77
	*MENINGITIS	02	01.53
ACCORDING TO YOU WHAT CAUSES GENETIC ABNORMALITY?	*UNNECESSARY EXPOSURE TO X RAY	13	10.00
	*LYSERGIC ACID DIETHYLAMIDE DRUG	18	13.85
	*ALCOHOL CONSUMPTION DURING PREGNENCY	07	05.38
	*ALL OF THESE	92	70.77
WHAT IS CAUSE OF	*RH POSITIVE MOTHER AND RH NEGATIVE FOETUS	37	28.46
ERYTHROBLASTOSIS FOETALIS	*RH NEGATIVE MOTHER AND RH POSITIVE FOETUS	64	49.23
?	*BOTH MOTHER AND FOETUS ARE RH POSITIVE	19	14.62
	*BOTH MOTHER AND FOETUS ARE RH NEGATIVE	10	07.69
	WHAT DO YOU THINK IS CAUSE OF GENETIC DISEASE? WHICH OF THE FOLLOWING DISEASE IS INHERITORY? ACCORDING TO YOU WHAT CAUSES GENETIC ABNORMALITY? WHAT IS CAUSE OF ERYTHROBLASTOSIS FOETALIS	DISEASE ? *DISEASES INHERITED FROM BIOLOGICAL FATHER AND MOTHER *DISEASE OF BLOOD *DISEASE RELATED TO SKIN WHAT DO YOU THINK IS CAUSE OF BLOOD *TOUR ARRIAGE *INTER CASTE MARRIAGE *INTER RELIGIOUS MARRIAGE *INTER RELIGIOUS MARRIAGE *ACCIDENT WHICH OF THE FOLLOWING DISEASE IS INHERITORY? *ALBINISM *VITAMIN DEFICIENCY *ANEMIA *MENINGITIS ACCORDING TO YOU WHAT CAUSES GENETIC ABNORMALITY? *ALCOHOL CONSUMPTION DURING PREGNENCY *ALCOHOL CONSUMPTION DURING PREGNENCY *ALL OF THESE WHAT IS CAUSE OF ERYTHROBLASTOSIS FOETALIS? *RH POSITIVE MOTHER AND RH POSITIVE FOETUS *BOTH MOTHER AND FOETUS ARE RH POSITIVE *BOTH MOTHER AND FOETUS ARE RH POSITIVE	WHAT IS MEANT BY GENETIC DISEASE? *DISEASES RELATED TO BONE *DISEASE SINHERITED FROM BIOLOGICAL FATHER AND MOTHER *DISEASE OF BLOOD *DISEASE OF BLOOD *CONSANGUINOUS MARRIAGE *INTER CASTE MARRIAGE *INTER RELIGIOUS MARRIAGE *INTER RELIGIOUS MARRIAGE *INTER RELIGIOUS MARRIAGE *ACCIDENT *ALBINISM *ALBINISM *ALBINISM *VITAMIN DEFICIENCY *ANEMIA *WENINGITIS ACCORDING TO YOU WHAT CAUSES GENETIC ABNORMALITY? **EYENGIC ACID DIETHYLAMIDE DRUG *ALCOHOL CONSUMPTION DURING PREGNENCY *ALCOHOL CONSUMPTION DURING PR

Continue

6.	WHAT IS HAEMOPHILIA ?	*HEREDITORY BLOOD DISORDER	107	82.31
		*HEREDITORY BONE DISORDER	05	03.85
		*BLOOD CANCER	07	05.38
		*HEREDITORY DISEASE AFFECTING MENTAL HEALTH	11	08.46
7.	WHAT IS DOWN SYNDROME?	*GENETIC DISORDER IN WHICH MENTAL AND PHYSICAL HEALTH IS	116	89.23
		AFFECTED		
		*CONGENITAL DISEASE OF KIDNEY	06	04.61
		*INFECTIOUS DISEASE OF BRAIN	03	02.31
NO	OTTECTIONS	*BONE DISORDER OPTIONS	05	03.85
NO	QUESTIONS	OPTIONS	FREQUENCY (n)=130	PERCENTAGE (%)
8.	WHAT IS SICKLE CELL ANEMIA ?	*INHERITED BLOOD DISEASE ABNORMAL HEMOGLOBIN	108	83.07
	ANDAIN.	*INHERITED KIDNEY DISEASE	06	04.61
		*INHERITED LUNG DISEASE	03	02.31
		*IRON DEFICIENCY ANEMIA	13	10.00
9.	ACCORDING TO YOU WHAT IS	*BLOOD DISEASE	106	81.53
	THALASEMIA ?	*DISEASE RELATED TO NERVOUS SYSTEM	14	10.76
		*KIDNEY DISEASE	04	03.08
		*SKIN DISEASE	06	04.61
10.	WHAT IS CYSTIC FIBROSIS ?	*INHERITED BLOOD DISORDER	31	23.85
10.		*INHERITED DISEASE AFFECTING RESPIRATORY AND GASTRO- INTESTINAL TRACTS	71	54.61
		*DISEASE OF URINARY BLADDER	11	08.46
		*DISEASE OF LIVER	17	13.08
11.	WHAT IS PHENYLKETONURIA ?	*INHERITED DISEASE RESULTING IN DEFICIENCY OF LIVER ENZYME	75	57.70
		*DISEASE CAUSED BY ACCIDENTAL CONSUMPTION OF UREA	15	11.53
		*DISEASE OF CARDIOVASCULAR SYSTEM	08	06.53
		*INHERITED KIDNEY DISEASE	32	24.61
12.	WHAT IS FOLLOWING IS TRUE	*MARRIAGE BETWEEN BLOOD RELATIVES	21	16.15
	ABOUT CONSANGUINOUS MARRIAGE ?	*MARRIAGE BETWEEN SON OR DAUGHTER OF UNCLE / AUNT	19	14.61
		*THERE IS INCREASED RISK OF GENETIC DISEASES IN THESE MARRIAGES	19	14.61
		*ALL THE ABOVE	71	54.62
13.	FREQUENCY OF CASES OF	*18-24 Years	24	18.46
	DOWN SYNDROME IS MORE IN	*25-30 Years	26	20.00
	WHICH AGE GROUP OF	*31-39 Years	38	29.23
	MOTHERS ?	*40 Years AND ABOVE	42	32.30
14.	DO YOU THINK LATE MARRIAGES INCRESES RISK OF GENETIC DISEASES ?	*YES *NO	88 88	32.30 67.69
		*COUNSELLING OF STUDENTS	07	05.38
15.	WHAT IS GENETIC	*COUNSELLING REGARDING FAMILY PLANNING	13	10.00
	COUNSELLING ?	*COUNSELLING REGARDING INHERITED GENETIC DISEASE	107	82.30
		*MENTAL HEALTH COUNSELLING	03	02.31
16.	WHICH DISEASES CAN BE	*THALASSEMIA	06	04.61
	SCREENED FOR GENETIC	*HAEMOPHILIA	07	05.38
	COUNSELLING ?	*ALBINISM	10	07.69
		*ALL THE ABOVE	107	82.30
17.	WOULD YOU LIKE TO SCREEN	*Yes ME ONLY	04	03.08
1/.	FOR GENETIC DISEASE	*YES BOTH ME AND MY PARTNER	111	85.38
	BEFORE MARRIAGE ?	*YES ONLY MY PARTNER	05	03.85
		*NO	10	07.69
			-	
18.	BEFORE MARRYING DO YOU INQUIRE ABOUT PRESENCE OF	*YES	93	71.53
	GENETIC DISEASE IN FAMILY OF PERSON YOU WISH TO MARRY?	*NO	37	28.46
19.	DO YOU CHECK BLOOD GROUP AND RH FACTOR OF	*YES	83	63.85
	PARTNER BEFORE MARRIAGE	*NO	47	36.15
20	? WHY WOULD YOU LIKE TO		-	
20.	SCREEN YOURSELF AND YOUR	*IT CAN REDUCE THE FREQUENCY OF GENETIC DISEASE	15	11.54
	PARTNER BEFORE MARRIAGE ?	*IT CAN REDUCE THE COST OF EXPENDITURE ON HOSPITALIZATION OF CHILD BORN WITH DEFECT	04	03.07
		*IT WILL IMPROVE QUALITY OF LIFE OF CHILD BORN WITHOUT GENETIC DISEASE	20	15.38
		*ALL THE ABOVE	91	70.00

hospitalization of child born with defect, it will improve quality of life of child born without genetic disease.

CONCLUSION AND WAY FORWARD

Although the participants had adequate knowledge of genetic diseases, there was a gap in between knowledge and practices. Although most of the participants were decently educated there knowledge and awareness about some genetic diseases is not satisfactory and reason could be their non-health science or non-biology educational background. Ministry of Health and Family Welfare, GoI along with support of State and local health department should create a monitoring system and should organize awareness campaign especially at schools, colleges and should take necessary steps to educate people regarding genetic diseases. Consanguineous marriages are common in India as both individuals and their families prefer to get married within blood lines but consanguineous marriage have drawbacks as both individuals share common ancestor which increases the chances of expression of recessive genes involving mental retardation and congenital malformations in the progeny so consanguineous marriages should be avoided. Some diseases like Sickle cell anemia, thalassemia, hemophilia are common in tribal population mainly due poor education due to traditions involving consanguineous marriage.3,4,5,11,14,15 Chromosomal disorders like Down's syndrome are seen to be more prevalent in female having pregnancy in her late 40s. The overall risk of giving birth to an infant with Down's syndrome is 1:800. The risk rises sharply with age; it is 1:67 for those aged 40-45. Excessive consumption of alcohol during pregnancy can lead to malformations in foetus.^{6,12} It is estimated that about 10000-15000 babies with Thalassemia Major (TM) are born every year. The only cure available for these children with thalassemia major is bone marrow transplantation (BMT) more appropriately called hematopoietic stem cell transplant (HSCT). However, this can help only a few patients because of cost, paucity of BMT centres, or non-availability of a suitable HLA matched donor. Therefore, the mainstay of treatment is a regimen of regular blood transfusions followed by adequately monitored iron chelation therapy to remove the excessive iron overload-as a consequence of the multiple blood transfusions. Thus it is a transfusion dependent disorder and places a great burden on healthcare services.^{9,10} Although Ministry of Health and Family Welfare, Government of India have issued guidelines regarding Prevention and control hemoglobinopathies in india-thalassemia, sickle cell disease and other variant hemoglobin, 2016 and NATIONAL POLICY ON RARE DISEASES, 2021. 9,10 Also, the Ministry of Tribal Affairs has launched the Sickle Cell Disease Support Corner to bridge the gap between patients and health care services in tribal areas. The Portal provides a web-based patient powered registration system which will collate all information related to Sickle Cell Disease among tribal people in India, The portal will give access to real time data to every visitor through a dashboard, online self-registration facility, and will act as a knowledge repository with information about the disease and various government initiatives. The National Council on Sickle Cell Disease has also been constituted of senior officials of GoI and health care private and public bodies for timely and effective action.8 Member of Parliament Dr ShrikantEknathShinde, has introduced THE FREE AND COMPULSORY PRE-MARITAL GENETIC TESTING BILL, 2019 draft bill in Lok Sabhato provide for free and compulsory pre-marital genetic testing for couples planning to get married or start a family in order to identify common

genetic blood disorders like sickle cell anemia and thalassemia and for matters connected therewith or incidental thereto. ⁷ But there is large gap between knowledge of people and practices followed, significant expenditure should be focused on preventive aspect rather than curative aspect. Educating students, parents, teachers about genetic diseases and their effects on progeny with the help of Non-governmental organizations will not only fasten but also smoothen the process of educating the population. Prevention of genetic diseases can save millions of rupees spent and can reduce mental-emotional and psychological agony faced by patients and their parents. India is the youngest country in the world, youngsters who are considered as an asset of the county, and has a major role to play in its development. But a youth with severe intellectual disabilities and other genetic abnormalities will further increase the burden on country's economy and thus emphasis should be laid on creating awareness about genetic disease among youth population through mass campaign. This Study provides data necessary for the formulation of policies that will help to ensure awareness of population about genetic diseases in the Akola city.

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