



## THE URGENCY OF GENETIC RESEARCH IN HUMANS

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### ABSTRACT

Genetic research is a breakthrough in the field of molecular biology which has been proven to contribute to health both in diagnosis and therapy. To support the achievement of these competencies, health students need to view the importance of genetic studies in humans. A person's understanding of a subject is greatly influenced by a person's cognition and the information they absorb. Using a survey approach to students and stakeholders of health science universities. The aim of this study to obtain an overview of health students' understanding of the urgency of genetic research in humans. The level of student understanding of the meaning of genetics is considered quite good with an average score of 2.10 on a scale of 0 – 3. Based on the assessment of various genetic research titles, students' understanding of the urgency of genetic research in humans is relatively high, with an average score of 3.19 on a scale of 1-4. There were no differences in student assessments regarding the urgency of genetic research in humans in each class. Significant differences based on generation appear in students' preferences for research on human genetics.

Keywords: genetics; human; molecular

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## INTRODUCTION

Genetic research on humans in Indonesia receives serious support from the Indonesian government. Through the Ministry of Health of the Republic of Indonesia, genetic research studies were confirmed with the inauguration of the Biomedical and Genome Science Initiative (BGSi) on August 14 2022. This institution aims to support diagnosis, care and treatment services in Indonesia using genome information (Kemenkes RI, 2023) (1). This genetic information is managed through technology known as Whole Genome Sequencing (WGS).

Genetic research in Indonesia, which has long been carried out by the Eijkman Institute, has received a new platform. Under the Ministry of Health, BGSi can concentrate genome services on 6 main types of disease by appointing 6 national hospitals. The six main diseases are cancer, infectious diseases, brain and neurodegenerative diseases, metabolic diseases, genetic disorders and aging, as well as diseases related to nutritional problems (Kemenkes, 2022) . Starting in 2022, BGSi itself is targeting 10 thousand human genome sequences in two years to map variants of the genome data of the Indonesian population who have these priority diseases.

The benefits of genetic research in humans are not only in the health sector. For Indonesia, which has racial diversity, this research is able to trace the origins of Indonesian people who are spread across various tribes and races. A study published in 2017 found the complexity of population history in Indonesia. Based on genome-scale data from 25 populations from various islands, the research found that western Indonesia has a more complicated mixing history than eastern Indonesia. This mixing was formed from interactions with new arrivals from mainland Asia and Austroonesia (Hudjashov et al., 2017). Research oriented towards racial diversity in Indonesia can be developed to see the susceptibility and resistance of certain ethnicities to genetic diseases (Kompas.com, 2015).

The existence of the Indonesian Ministry of Health in genetic research in humans can be an inspiration for the academic world in the health sector to get involved in this movement. One form of concrete involvement is including molecular biology studies in the learning curriculum. The impact is not only an increase in the number of genetic research on humans, but also on students' concerns about the research itself. Based on the explanation above, genetic research in humans still opens up broad opportunities for improving health services in Indonesia. For this reason, health students need to understand the urgency of this field of study. For study programs/departments that include biomolecular studies in their curriculum, it is necessary to measure students' understanding of the urgency of genetic research in humans.

## **METHOD**

This research took place at the National College of Health Sciences. This type of research is quantitative research, namely research that tries to describe a problem whose results can be generalized (Eka Kartika Untari, 2022). The method used is a survey method of respondents to determine the description of a phenomenon. The instrument used is a direct questionnaire to respondents to obtain information about themselves and things they know (Arikunto, 2013). Respondents were active regular class students in medical laboratory technology study programs, both diploma three and applied bachelor degrees. The total population is 300 students. Samples were taken using a proportional stratified technique. This technique groups populations into categories called strata (Arikunto, 2013). The strata are determined based on the class in each generation for each study program. This was done considering that each strata group has relatively the same degree of intensity and interaction regarding genetic research topics on humans. The number of samples in each group follows the Arkin and Colton table, namely 50%. This number has a confidence level of 95% and a standard error of +5% (Slamet, 2001).

Health Students' Understanding of the Urgency of Genetic Research in Humans is the dependent variable in this research. This variable will describe the high or low degree of the respondent's understanding of the urgency of genetic research in humans. Meanwhile, the independent variable is the group of respondents, as a group that has a certain level of intensity of exposure to the theme of genetic research in humans. In this way, this research can provide an overview of the differences in understanding between each group regarding the urgency of genetic research in humans. The work procedures for this research include distributing questionnaires, collecting questionnaires, determining research data, and analyzing research data. Questionnaires were collected directly from respondents. Next, the completed questionnaires will be collected based on respondent groups. Each questionnaire will be checked on an informed consent sheet, and respondents who give consent will continue with the process of determining research data. By using the Excel application or online randomizer, questionnaires for each class will be selected as research data according to

the number that has been determined proportionally. The data will then be given a numerical code on an ordinal scale for each research variable. This codified data will be processed using statistical tests.

Data analysis includes validity tests, reliability tests, and difference tests to see differences in understanding in each group. Validity tests were carried out on 20 samples using product moment correlation. Reliability testing is carried out to ensure the consistency of measuring instruments. Measuring instruments are said to be reliable if they are stable, reliable and constant. (16). To measure this reliability, Cronbach's alpha ( $\alpha$ ) coefficient formulation is used. Meanwhile, differences in the level of understanding in each group will be measured using the Kruskal Wallis test. Both validity, reliability and difference tests in this research will be carried out using the SPSS 20 application

## RESULTS

Apart from using students as respondents, the urgency of genetic research in humans is also seeking responses from stakeholders in health education institutions. There were 154 student respondents, while there were 35 respondents from stakeholders. The following is the distribution of respondents from student groups and stake holders:

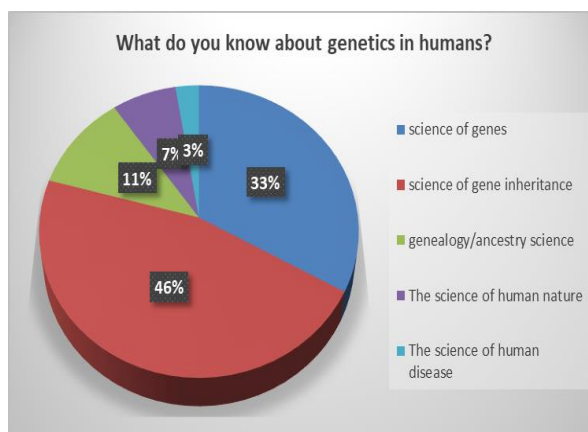
Table 1.  
Distribution of Student Respondents

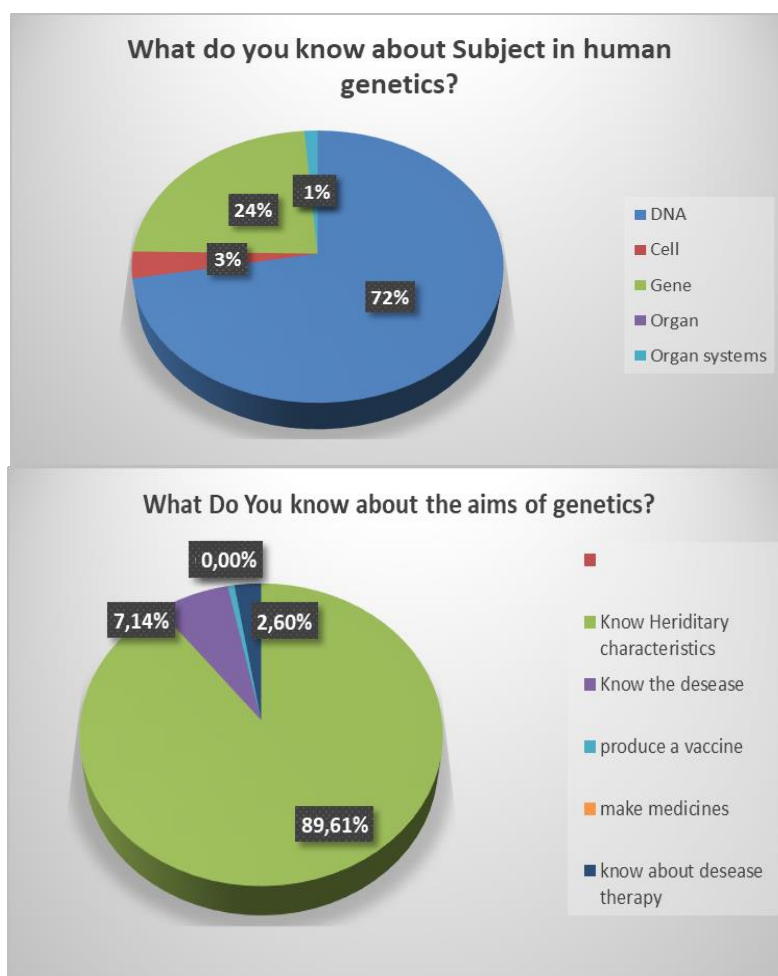
Division	Years of entrance		
	21/22	22/23	23/24
DIII TLM	50	11	10
STR TLM	33	25	25
	83	36	35

Table 2.  
Distribution of Stakeholder Respondents

Institution	Amount
Clinical Lab	6
Hospital/Public Health Center	25
Pharmaceutical Industry	1
Other Healthcare Industries	3
	35

To determine health students' understanding of the urgency of genetic research in humans, first, students' understanding of the concept of genetics was measured. There are three basic questions regarding the concept of genetics, namely: What do you know about human genetics? What do you know about material in human genetics? and What do you know about the aims of genetics?. An illustration of students' understanding of genetics can be seen in the following diagram:





The correct answer to the first question was 46.1%, 74% to the second question, and 89.6% to the third question. If the scores of each respondent on the three questions are combined, then there are 2 respondents with a score of 0, 34 respondents with a score of 1, 68 respondents with a score of 2, and 49 respondents with the highest score, namely 3. Descriptive statistics if the scores of each respondent are combined are as follows:

Table 3.  
Descriptive statistics if the scores of each respondent

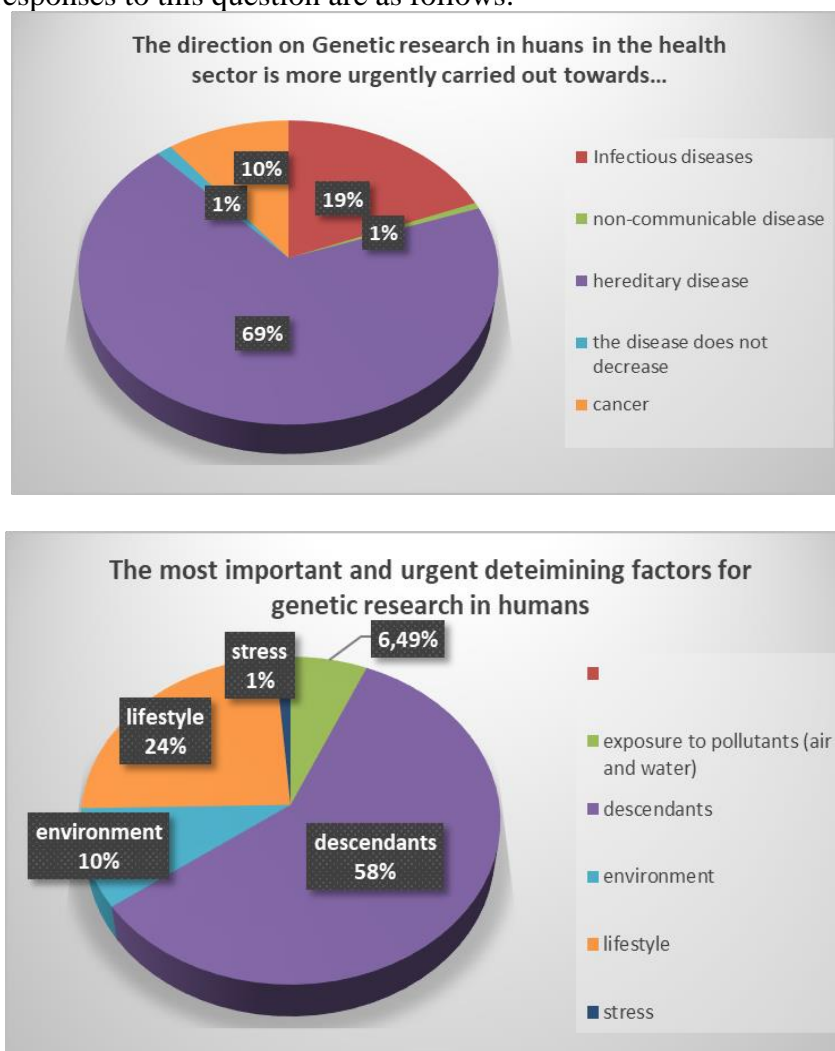
Descriptive Statistics					
	N	Minimum	Maximum	Mean	Std. Deviation
Understanding	145	.00	3.00	2.1034	.77032
Valid N (listwise)	145				

Is this understanding influenced by the length of study indicated by the student's year of entry? The following Kruskal Wallis test statistical test will prove the difference in understanding between the classes, namely the classes of 2020, 2021, 2022, and 2023.

Ranks			
	Class	N	Mean Rank
Understanding	1	16	66.75
	2	64	78.92
	3	31	71.95
	4	33	63.35
	Total	144	

Test Statistics <sup>a,b</sup>	
	Pemahaman
Chi-Square	3.942
df	3
Asymp. Sig.	.268

Even though there are differences in the average ranking of understanding of genetics in each class, the statistical test shows a p value (Asymp.Sig) of 0.266 or greater than 0.05. Thus, the difference in understanding of genetics based on generation is not significant at the p value: 0.268. After knowing the respondent's level of understanding of the concept of genetics, questions were then asked related to the urgency of the direction of genetic research on health, as well as understanding the determining factors of genetic research. The results of respondents' responses to this question are as follows:



In the final section, respondents from the student group were given several research titles on the topics Diabetes mellitus and Tuberculosis. These two topics cover the fields of Clinical Chemistry, Hematology, Bacteriology, Parasitology as a comparison and Molecular Biology where the study of genetics is studied. All respondents were asked to rate each title submitted, based on the ratings: (1) Not important and not urgent, (2) Not important but urgent (3) important but not urgent, and (4) important and urgent. The comparison of the average responses of respondents to the research titles is as follows:

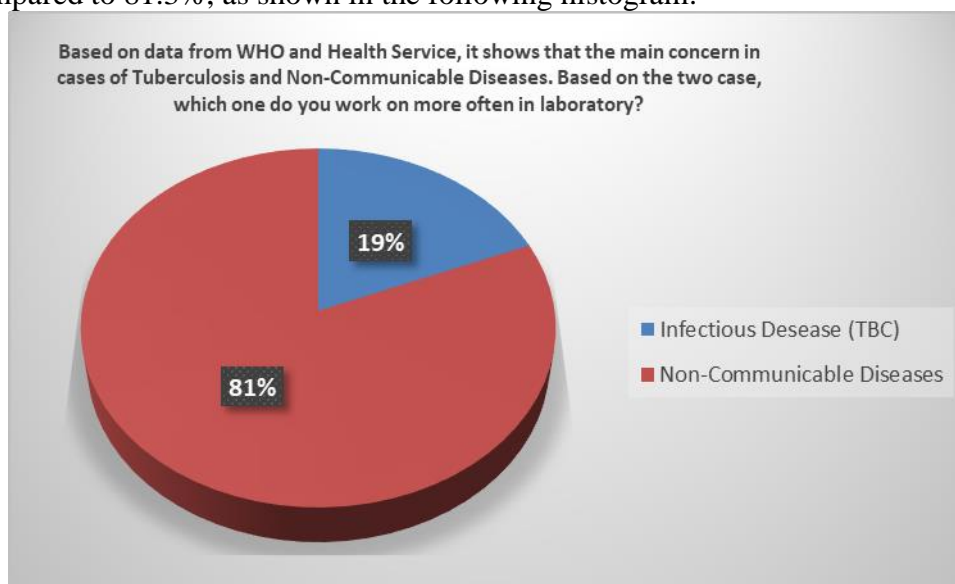
Table 4.

The comparison of the average responses of respondents to the research titles

TOPIC	Biomol	K. Klinik	Hematology	Bakteriology	Parasitology
Diabetes Melitus	3,24	3,03	3,18	2,78	3,05
Tuberculosis	3,12	3,23	3,17	3,12	3,28

The table above shows that the field of study of Molecular Biology has a higher average ranking on the topic of Diabetes mellitus than Tuberculosis. This is in accordance with a

survey of stakeholders in the field of clinical laboratories and hospitals, that TB disease actually occupies a smaller percentage than non-communicable diseases with a comparison of 18.8% compared to 81.3%, as shown in the following histogram:



The differences in student assessments based on class (year of entry) in terms of the level of urgency regarding student research titles can be seen in the following table:

Ranks			Test Statistics <sup>a,b</sup>	
Class	N	Mean Rank		Total Gen
1	16	57.41	Chi-Square	4.604
2	64	69.52	df	3
urgency 3	31	75.98	Asymp. Sig.	.203
4	33	82.33		
Total	144			

The average ranking for each class shows that the younger the class, the higher the average score, or the more they give a high level of urgency to biomolecular research titles. The difference in student assessments of research titles in the biomolecular field by each generation shows a value of  $p = 0.203$  or much greater than the  $p$  table (0.05). Thus, there are no significant differences in the assessment of the level of urgency for genetic research in each generation. Furthermore, students' understanding of the urgency of genetic research will be seen based on differences in student preferences for genetic research in humans, as well as differences in student assessments of biomolecular research titles. Based on statistical tests on student preferences for genetic research in humans, the following data were obtained:

Ranks			Test Statistics <sup>a,b</sup>	
Class	N	Mean Rank		Preference
1	16	87.44	Chi-Square	16.479
2	64	57.34	df	3
Preference 3	31	78.55	Asymp. Sig.	.001
4	33	88.97		
Total	144			

The difference in student preferences for genetic research based on generation shows a value

of  $p = 0.001$  or much smaller than the  $p$  table (0.05). Thus, there are significant differences in preferences for student research in each generation.

## **DISCUSSION**

Research into human genetics underwent a major revolution at the end of the 19th century. Matt Redly (1993) summarizes various studies of genetic research throughout the world. Several things related to the health sector include information about disease, healing and prevention recorded in chromosomes 9, 18 and 19 (Ridley, 1999). Indonesia's racial diversity influences an individual's genetic code. The differences in the physical and social environment in Indonesia have inspired various research into human genetics, especially in the health sector. Research on the whole blood microbiome of several regions in Indonesia has found that differences in geographical environment have an impact on the presence of certain types of pathogens. The presence of plasmodium and flavivirus infections is found mostly on the easternmost island of Indonesia (Susilowati et al., 2022). This research clarifies the picture of data from the Indonesian Ministry of Health for 2021 that Papua and West Papua are the lowest ranking regions as areas eliminated from endemic malaria (Kemenkes RI, 2022). The existence of malaria infection in malaria patients in Indonesia is an interesting study. Bobowik, et al (2021) have discovered unique genes involved in RNA processing, splicing, and receptor genes in malaria cases in Indonesia. When compared with transcriptomic studies in Africa and America it was found that many of the pathways are the same in the recognition of receptors and genes related to inflammation, as well as innate immunity and cytokines in plasmodium infections (Bobowik et al., 2021). Penelitian pada penyakit tidak menular seperti Osteoarthritis (OA) yang merupakan penyakit sendi kronis paling umum juga telah menjadi topik dalam penelitian genetika pada manusia. Budhiparama, dkk (2023) menemukan gen polimorfisme yang memungkinkan menjadi faktor predisposisi OA lutut pada pria maupun wanita. Pada wanita dipengaruhi oleh keberadaan alel A SNP rs871659, sedangkan pada pria dipengaruhi oleh alel G SNP rs3771202 (Budhiparama et al., 2023).

Studies regarding understanding the importance of genetic research have been carried out, especially in science academics. Starting from an initial understanding of the concept of genetics for pupils, students, teachers and lecturers. This research shows that there is no difference in the concept of genetics for teachers, lecturers and students. For students, this initial understanding influences understanding of genetic concepts in genetics lectures (Nusantari, 2018). Another survey regarding understanding of genetics among biology study program students was conducted as a form of academic evaluation of genetics lectures. The survey shows that 50% of students do not understand genetics in a basic and independent manner (Nusantari, 2018).

A survey of understanding of genetics was also carried out because it considers genetics as a biological material which is considered difficult because the material is typically complicated and abstract. When lectures are carried out online, it is deemed necessary to measure the level of learning effectiveness in genetics courses (Prastyaningtias, 2022). Adhering to the same principle, studies on the understanding of genetics have also been carried out by measuring students' misconceptions about the concept of genetics itself (Wulandari et al., 2021). All of the surveys on student understanding of genetics above used student subjects in science study programs from various universities. For health study program students, measurements of understanding related to genetic research have been carried out, especially regarding pharmacogenomics. Participants consisting of medical, midwifery, pharmacy, nursing, nutrition and other students were observed to determine the level of knowledge, attitudes and perceptions regarding pharmacogenomics (Eka Kartika Untari, 2022).



The development of genetic research in recent years has given rise to increasing hope that findings in genetic research can provide information to improve health, predict the risk of disease, accelerate the development of new treatments and therapeutic techniques as well as answer population origins, migration patterns, or the influence of epigenetic factors related to the spread of disease. (Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research, 2022) The following ethical principles need to be considered to answer the challenges of human genetic research, namely: reciprocity, mutuality, solidarity, citizenship, and universality. Reciprocity, namely that samples given often do not provide direct benefits to the giver. Mutuality means that not all predictive genetic tests are needed so they should only test according to the information needed. Solidarity is the lack of freedom to refuse to be a subject for reasons of benefiting the ethnicity/nation/state. Citizenship is related to public surveys and equal distribution of understanding. Universality is universality as the basis for genome characteristics (Anurogo & Parikesit, 2021)

Molecular genetics makes it possible to assess DNA variations directly. Advances in DNA microarray technology enable genome-wide association studies. This study succeeded in identifying several DNA variants that contribute to the heritability of behavioral traits. The ability to combine the effects of thousands of DNA variants in polygenic scores has created a DNA revolution in behavioral science that allows the use of DNA to predict individual behavioral differences (Plomin, 2023)

Genome-wide association studies provide a powerful way to identify genetic variants that play a role in common diseases. Studies of this kind present important ethical challenges (Vries et al, 2011). Genetic counseling is a comprehensive form of intervention compared to routine interventions or standard care such as psychoeducation (Mwangi & Mwayo, 2020) Genetic Counseling is used because it can help a person understand and adapt to the medical, psychological, family implications and genetic contributions to a disease that are not available in other interventions (Setiawan, Ediati, & Winarni, 2017). In Indonesia, research on genetic counseling interventions on acceptance of genetic diseases is still lacking, even patient awareness of receiving disease information is still low (Ariani, Soeharso, & Sjarif, 2017). In several developed countries such as the United States, Australia and the Netherlands, genetic counseling is carried out as a permanent procedure to treat psychological problems in people who have genetic diseases (Voils et al., 2015). Although there is increasing research on genetic counseling and its benefits, genetic counseling has not been widely implemented by health workers. In addition, the lack of knowledge or information about genetic diseases among lay people and health practitioners hinders the genetic counseling process (Ariyanto et al, 2021)

## **CONCLUSION**

The level of student understanding of the meaning of genetics is considered quite good with an average score of 2.10 on a scale of 0 – 3. Based on the assessment of various genetic research titles, students' understanding of the urgency of genetic research in humans is relatively high, with an average score of 3.19 on a scale of 1-4. There were no differences in student assessments regarding the urgency of genetic research in humans in each class. Significant differences based on generation appear in students' preferences for research on human genetics. Other statistical tests are needed to see the relationship between variables that determine students' understanding of the urgency of genetic research in humans with more indicators to measure students' basic knowledge, especially in the health sector who study molecular biology as well as a greater amount of research in the field of genetics, which



is appropriate to the level subject (respondent) education so that it can be compared with research from other fields of science.

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