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Analysis of Genetic Hearing Loss Using Automata Theory as a Framework

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Abstract

In this paper, we will refer to hereditary hearing loss. Hearing loss is a genetic as well as accidental problem. Genetic deafness is a type of hearing loss that is caused by changes or mutations in genes related to the hearing process. These mutations can be inherited from one or both parents and can occur spontaneously. There are two types of genetic deafness: Syndromic and non-syndromic. The inheritance of genetic deafness depends on the type of mutation and the pattern of inheritance. Some mutations are dominant, other mutations are recessive, X-linked inheritance is another type of inheritance, and mitochondrial inheritance which are described in detail in our paper. Some authors have suggested that rather than publicly condemning deaf parents that choose deafness for their children, we believe that this technique can be viewed as unacceptable. In today's technological world it is needed to construct and design models related to certain topics and automate it. The idea of how to create an automata model with some techniques is described in this paper. But deaf people are still considered less capable as compared to normal humans, which do not help in the betterment of the society but rather to select genetic factors that are typically seen as a handicap by majority of the sociological environment. The inheritance of genetic deafness follows the rules of Mendelian genetics, where the genes are passed down from parents to their offspring.

Keywords: Hearing loss • Genetic algorithm • Hierarchy of genetic algorithm • Automata • Finite state machine • Genetics

Introduction

In this paper, we proposed the idea of how to use automata in genetic algorithm for Hereditary Hearing Loss (HHL) and how it will help us in future use for Artificial Intelligence (AI). The focus of this paper is on how the person inherited hearing loss disorder through parental side [1].

Genetic algorithm for hearing loss

Genetic algorithms are a type of optimization algorithm that is inspired by natural evolution. They can be used to identify patterns in data and generate hypotheses about the underlying causes of a condition. The steps to solve GA begins with the optimization of GA and then generating random population known as encoding used to generate initial population. Apply fitness function F(x) to calculate the fitness of best chromosome. Select a pair of best chromes to form offspring. If we find the best offspring, then the pairing is completed, and we can end the optimization process but if the pairing of chromosome is not best or are less fit than following pair. So, we replace the old population and end the optimization process [2].

Use in AI

Genetic Algorithms (GAs) are a type of optimization algorithm that can be used in Artificial Intelligence (AI) and machine learning to find the optimal solution to a problem. They are particularly useful for solving problems that involve many variables or have complex, non-linear relationships. GAs can be used to optimize the performance of a model by searching through a large space of practical solutions and selecting the best ones based on some measure of fitness. In AI and machine learning, GAs is often used to optimize the weights of a neural network or select the most effective features for a machine learning model [3].

GA work in automata

Genetic Algorithms (GAs) can be used to optimize the transitions between states in an automaton, a mathematical model that represents the behavior of a system. GAs search through a large space of transitions, selecting the best ones based on some measure of fitness to achieve a desired objective. They can be useful for optimizing complex, non-linear systems and finding solutions that trade off multiple objectives [4].

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Genetic flow of hearing loss

Hearing loss: Hearing is a complex process so the causes of hearing loss are also complex and can be due to damage to the inner ear, genetic factors, environmental factors, or a combination of these. Understanding the genetic causes of deafness is important as it can inform families about their chances of having children with hearing loss and can also impact treatment options. Genetic deafness can be passed on to future generations, even if neither parent is affected. Genetic testing can help in understanding the genetics of hearing loss, but it can be complicated and difficult to understand, so it is recommended to consult with a physician, genetic counselor, or clinical geneticist for more information [5].

How is hearing loss detected and diagnosed?

The early detection of hearing loss is crucial for a child's communication and learning skills, which is why many states now provide a simple hearing test for newborns. Without this screening, hearing loss may go unnoticed until the child has difficulties speaking and learning, which could be as late as 2-3 years of age. Babies who do not pass the screening are referred to an audiologist, who will determine the severity and type of hearing loss. Hearing loss severity is measured by the hearing threshold level, and hearing loss types are classified according to which part of the hearing system is affected. Conductive hearing loss occurs in the outer and middle ear, while sensor neural hearing loss occurs in the inner ear [6].

When a child is diagnosed with hearing loss, further evaluation is needed to determine the cause. The cause could be due to things like loud noise, head injury, medications, infections, or genetic factors. Genetic deafness can be syndromic or nonsyndromic and a detailed family history can help identify the cause. However, absence of a family history of deafness does not rule out the possibility of genetic deafness. Special tests may be needed to diagnose certain syndromes that include deafness as a feature, and other specialists may be involved in the evaluation process. Identifying the cause of hearing loss can help with treatment, management, and predicting the possibility of passing on deafness to future generations [7].

Literature Review

How is hearing loss inherited?

Half of childhood deafness is due to hereditary causes involving genes in the hearing process that are inherited or passed down in a family. Genes are made of DNA sequences and humans have about 100,000 different genes grouped into small structures called chromosomes. People have twenty-three pairs of chromosomes, including a pair of sex chromosomes, and each pair consists of one chromosome inherited from the mother and another from the father. Gene mutations can be dominant or recessive, and carriers of recessive mutations can pass them on to their children. X-linked inheritance involves recessive mutations in genes on the X chromosome, while mitochondrial inheritance can occur due to mutations in the mitochondrial DNA [8].

The article also provides an example of a mutation in a gene associated with hearing and how it can alter the instructions contained in the DNA sequence.

An example of a mutation in a gene associated with hearing where a single base change from G to T is enough to alter the instructions contained in the DNA sequence (Figure 1). The normal sequence is AGATGAGCA, while the mutated sequence is AGATTAGCA, resulting in a non-working gene. This illustrates how mutations can disrupt the gene enough so that it does not function correctly and can lead to hearing loss [9].

... A G A T G A G C A... normal sequence=working gene

... A G A T T A G C A... mutated sequence=non-working gene

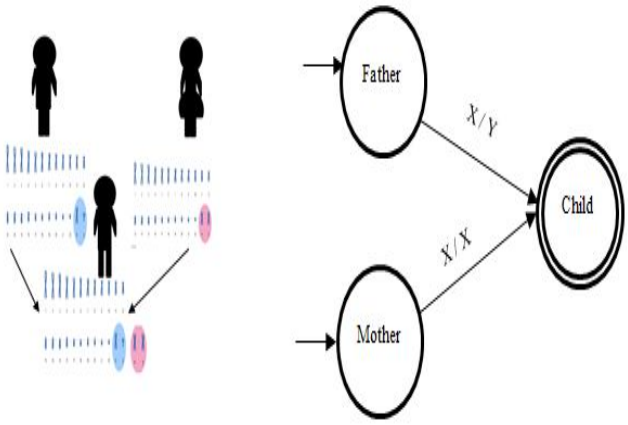

Figure 1. Each pair of chromosomes in a person's twenty-three pairs, which also include a pair of sex chromosomes, is made up of one chromosome inherited from the mother and one from the father. Gene mutations can be recessive or dominant, and carriers of the latter type of mutation can pass it on to their offspring.

Inheritance of a dominant mutation

Inheritable genetic information is passed down from parents to their children through chromosomes. A child receives one copy of each chromosome from their mother (colored green) and one from their father (colored blue). If a dominant mutation is present on one copy of the father's chromosomes, it is represented by a red band. As the mother has two unaltered copies of the chromosomes, all her offspring will receive an unaltered copy from her. However, each child has a 50% chance of inheriting the copy with the dominant mutation from their father (Figure 2). This means that in each pregnancy, there is a 50% chance that the child will inherit the dominant mutation and be affected; as only one copy of the mutation is required for it to be expressed dominantly [10].

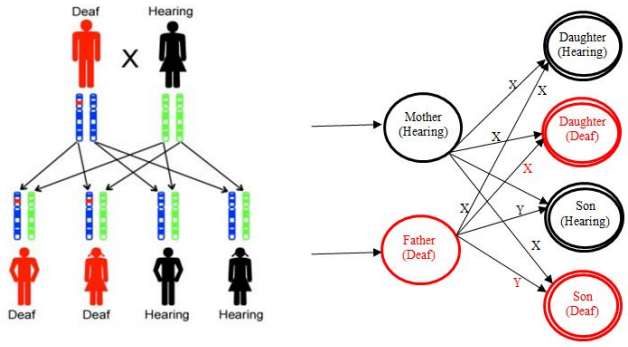

Figure 2. Each child has a 50% chance of receiving the father's copy of the dominant mutation. As just one copy of the mutation is necessary for it to be expressed dominantly, this indicates that there is a 50% probability that the child will inherit the dominant mutation and be impacted in every pregnancy.

Inheritance of a recessive mutation

The inheritance of chromosomes in humans follows a pattern where each child receives one copy of a chromosome from their mother (shown in green) and one copy from their father (shown in blue). If there is a recessive mutation in a gene on one copy of a parent's chromosome, represented by a red band, the child has a 50% chance of inheriting that mutated copy from either parent. However, for a recessive mutation to have an effect, both copies of the gene need to be mutated. To calculate the probability of two independent events occurring simultaneously, one needs to multiply the probabilities of each event occurring separately (Figure 3).

It can be stated that in every pregnancy, there is a 25%likelihood that the child will acquire both mutations, resulting in one out of four children being born deaf on average [11].

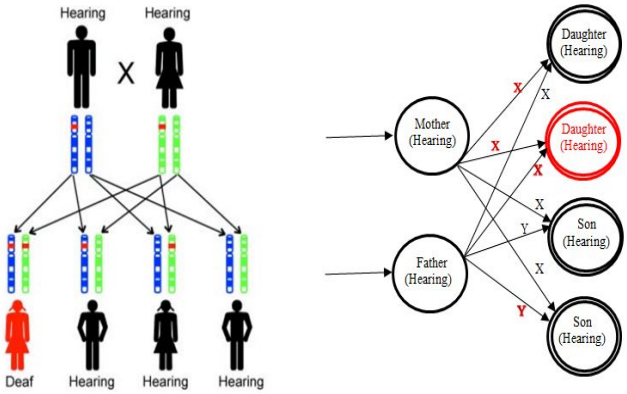

Figure 3. Most of the genetic hearing loss is up to seventy-five percentage of inheriting chromosomes from recessive side that is; up to twenty percent of genes are attributable to dominant genes. It can be said that there is a 25% chance that both mutations will occur during a pregnancy, leading to an average of one out of four infants being born deaf.

Inheritance of an X-linked recessive mutation

Children inherit one sex chromosome from each of their parents, with girls inheriting two X chromosomes from their mother and father, while boys inherit an X chromosome from their mother and a Y chromosome from their father. If a recessive mutation is present on one of the mother's X chromosomes, girls can remain unaffected as they will inherit an unaltered X chromosome from their father. However, boys do not have another X chromosome to compensate for the mutated one, and therefore have a 50% chance of inheriting the altered X chromosome from their mother, which can result in deafness [12].

Inheritance of a mitochondrial mutation

During reproduction, only the mother's egg contributes mitochondria to the developing child, meaning that only females can pass on mitochondrial traits to their offspring. If the mother has a mitochondrial gene mutation, she will pass it on to all her children, while males cannot pass on mitochondrial mutations. However, spontaneous mutations can occur when a gene in a parent's egg or sperm cell undergoes a DNA change, resulting in a genetic trait suddenly appearing in a family without a previous history of it. The chance of future generations inheriting deafness can be predicted in such cases.

While some forms of hearing loss are caused by mutations in single genes, others require mutations in two or more genes for a person to be affected (Figure 4). Additionally, mutations in certain genes may not directly cause hearing loss, but instead increase the risk of developing it due to environmental factors such as exposure to loud noise or antibiotics. Further research on individuals with hearing loss is necessary to fully understand these complex connections between genetics and deafness [13].

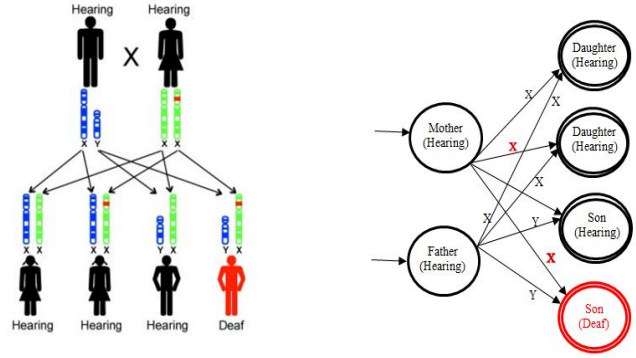

Figure 4. A little number of geneses of X-linked disorder. Boys have a 50% chance of receiving the changed X chromosome from their mother, which might cause deafness, because they lack a second X chromosome to make up for the defective one.

What is genetic testing?

Genetic testing is an essential tool to detect genetic mutations that could cause disorders such as hearing loss. However, it is important to note that genetic testing is only useful if the gene responsible for the condition is known. This is because a comparison between a particular person's gene sequence and that of the regularly occurring gene is necessary to detect mutations that could stop the gene from functioning [14].

In the case of deafness, mutations in the connexin twenty-six gene are the most common genetic cause of the condition, accounting for

up to half of recessive nonsyndromic hearing loss cases [15]. The connexin twenty-six gene tests is a widely available genetic test for hearing loss since the gene is short, making it easy to analyze. Therefore, any child born with hearing loss or developing it after birth is a candidate for the connexin twenty-six genetic test, especially in cases where no obvious causes of hearing loss are found.

It is worth noting that certain connexin twenty-six mutations are more prevalent in specific populations, such as the "35 delG" mutation commonly found in Caucasians. This mutation involves the deletion of the G at position thirty-five of the sequence. As more genes are discovered, genetic testing for various disorders will become more widely available, leading to more accurate diagnosis and treatment.

35delG mutation: The G at position thirty-five is highlighted in blue.

...C T G G G G G G T G T G A A C A A A C A C... hearing
...C T G G G G G V T G T G A A C A A A C A C... deaf

The connexin twenty-six gene is responsible for about half of recessive nonsyndromic hearing loss cases and mutations in this gene can be detected through genetic testing. Some connexin twenty-six mutations are more common in certain populations, such as 35delG in Caucasians and 167delT in Ashkenazi Jews. However, genetic testing results can be complicated and not always easy to interpret, as not all sequence changes in the gene cause hearing loss, and some people with hearing loss have mutations in other genes or non-genetic causes. As new genes contributing to hereditary hearing loss are discovered, the number of genetic tests for hearing loss will continue to increase [16].

How is genetic testing helpful?

Knowing the genetic cause of a person's hearing loss can help in making improved decisions about treatment and management, and predicting whether the hearing loss will worsen over time. It can also help determine what kind of damage has happened to the hearing system, and if problems besides hearing loss may be present or may develop in the future. Genetic testing can provide individuals or parents of a deaf child with the satisfaction of understanding the cause of the hearing loss and help in making reproductive choices. However, it can also be an overwhelming and stressful experience to learn that a mutation in one's own genes is the cause of their child's deafness. Genetic mutations are common, and no person is responsible for the genes they possess. Genetic counselors can help in educating and discussing the benefits and drawbacks of genetic testing and associated issues. It is important to understand these factors before making an informed decision about testing [17].

Over the last decade, an increasing number of automated approaches have already mentioned accuracy, reliability, as well as time efficiency comparable to manual hearing assessments. New developments, such as machine learning approaches, focus on manual audiometry in terms of features, versatility, and cost-effectiveness. Automated assessment methods using digital devices, when used within known constraints, can support task movement, self-care, telehealth, and patient care pathways. Hearing loss diagnosis and management require genetic testing can also be done to assist in the diagnosis of hearing loss, particularly when the origin of the hearing loss is unknown, or the hearing loss is hereditary. Genetic testing can detect genetic abnormalities that cause hearing loss, allowing the kind and degree of the hearing loss to be determined and treatment recommendations to be made. Hearing loss is often managed using a combination of medical and rehabilitative techniques. Our understanding of hearing loss genetics has aided in the development of focused medicines for certain forms of hearing loss, such as gene therapies and pharmacological treatments that target specific genetic pathways implicated in hearing loss. It has also aided in the selection of suitable therapies and rehabilitation strategies based on the hereditary etiology of hearing loss [18].

Discussion

In this study, we utilize data that will help us in future designing of an automata machine for hearing loss. This data is a subset of the Audio logical and Genetic Database (AudGenDG). The dataset has 53586 rows and six columns. Microsoft Excel is used to create pattern that help us to in the creation of a finite state machine to ascertain author's point of view on hereditary hearing loss that a person is deaf from left ear or right ear in addition with their ages, gender, frequency of hearing loss and ethnicity [19]. The user is provided with the idea of how they can use this machine to analyze the hearing range based on certain parameters. It also could show how the machine works in the inheritance of deafness from gender to patient ear. The dataset is visualized with the help of graphical representation (patterns) of certain parameters of dataset. Techniques we use certain Finite state machines with output like Mealy and Moore machines. These machines are used to determine the current state as well as the output value. Finite machines are the mathematical models or machines of any system that have the same input as well as output depending on certain situation with some limited conditional states [20].

Offered model

The data of deaf patients according to the parental side from where the disease is transfer and after how many generations it is transfer is still very rare. So, in future it will be try to collect the data of generation gap and parental side too. This will help the user in identifying the exact generation gap of the patient to make the data available with one click. Otherwise, the full history of the patient will not be available. Following are the parameters of the design model (Figure 5).

HL: Hearing Loss; FH: Family History; X': Defected chromosome; X: Normal chromosome; Y: Normal chromosome; Father: Deaf; Mother: Normal; Output: 2 deaf child, 2 normal Child; N: Normal; D: Deafness

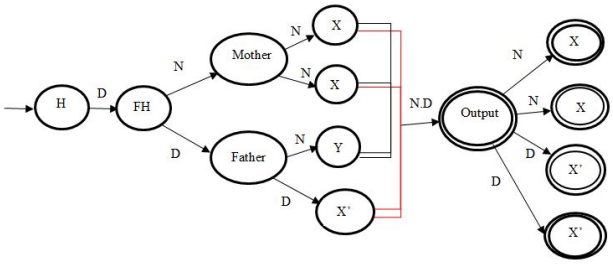

Figure 5. Parameters of offered model.

Working steps of offered model

In general, the finite automate we want to create is 'hereditary hearing loss.' Users will store the patient data using age and ethnicity to determine the similar data, different gender, unique diagnosis, and their preference.

Input variable(s): Age, frequency, gender, ethnicity, threshold unique. Diagnosis, pef Output variable: patient.

Ear state(s): Age, frequency, gender, ethnicity, unique. Diagnosis, pef, threshold Transition function: This would depend on the specific analysis you are conducting and the relationships you are interested in exploring between the input and output variables and the states.

Creating parameter

Gender: Where the user will store the ratio of different genders according to their unique diagnosis and the race from where that patient belongs.

Unique diagnosis: Where the data is stored according to their diagnosis that whether a person have deafness in left ear or right ear for helping in the creation of finite machine that also generates the output result of certain input values. Additionally, we use the dataset derived from Electronic Health Records (EHR) from the website figshare.com. AudGenDB is intended to be a public data source that represents a population enriched for hearing loss research. In this study we make the use of information from a survey that stores the patient information according to their gender. The finite automata or finite state machine were designed.

Parameters: Pattern for finding the HHL based on age, gender, family history of hearing loss, and severity of hearing loss. Suppose you have a dataset that includes information on one thousand individuals, including their age, gender, family history of hearing loss, and severity of hearing loss. You want to use this dataset to identify patterns of hereditary hearing loss.

Creation of finite machine: To do this, we could build a finite state machine that represents the different variables in our dataset. For example, we might define states for different age ranges, genders, and frequencies of hearing loss, and transitions between those states that represent the progression of the condition over time.

After creation of FM: Once we have built our automaton, we can use it to analyze the patterns of hereditary hearing loss in your dataset. For example, we might use simulations to examine how the severity of hearing loss changes over time for distinct groups of individuals, or we might use algorithms to identify clusters of individuals with similar characteristics.

Analyzing: By analyzing the automaton, we can uncover patterns in the data that suggest certain genetic or environmental factors are associated with an increased risk of hereditary hearing loss. For example, we might find that individuals with a family history of hearing loss are more likely to develop the condition, or that certain genetic mutations are associated with a higher risk of hearing loss (Figure 6).

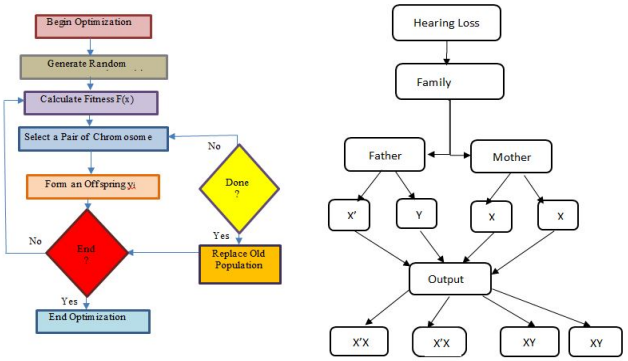

Figure 6. Parameters for finding the HHL.

Prominent feature of model

There are some signs for hearing loss such as hearing someone clearly and often misunderstanding what they say especially in noisy or crowded areas. Asking for people to repeat themselves repeatedly, listen to music, watch television with higher volume compared to normal people, and have difficulty hearing on the phone. The three types of models of deafness are based on either social or biological sciences. There are some cultural models for deafness such as the social model, and the infirmity or medical model. Our 'offered automata model' is based upon on genetic inheritance of hearing disability that is either inherited from one's parents, grandparents, our model can help those who have deafness or hearing disability from birth which is the result of inheriting defected chromosomes through DNA. By using this automata model one can easily understand another person's sayings, listen to music, watch television at normal volume, can easily be able to deal with phone calls and can communicate with others without any sort of difficulty. Our model can only help when a person is wearing the device created by using the abovementioned parameters and finite automata model. Also, the creation and wearing of the device depends on certain parameters and precautions of how to wear it and most importantly the steps to keep the device clean to avoid any sort of disturbance and infection in a person's ear.

Conclusion

By interpreting the results and drawing conclusions about the patterns of hereditary hearing loss in the dataset, you can contribute to a better understanding of the condition and potentially identify

innovative approaches for preventing or treating it. You may contribute to a better knowledge of the problem and find novel methods for prevention or treatment by evaluating the findings and making conclusions about the patterns of hereditary hearing loss in the dataset. Based on the observation of the result of patterns in hereditary hearing loss, we have concluded that mutations in chromosomes play the most significant role in determining whether deafness occurs or not in the genes. Genetic testing is an essential tool that helps in detecting genetic deafness. Through this technique we can identify whether genes are responsible for this or not. But the interpretation of genetic testing results can be challenging because not all gene sequence alterations lead to hearing loss and some individuals with hearing impairment may have mutations in genes other than those being evaluated or non-genetic causes. Moreover, with the discovery of new genes linked to hereditary hearing loss, the variety of genetic tests for this condition is expected to expand. So always keep this in mind while using this technique.

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