SATYA Special SCHOOL

RIST
RURAL INDIA SUPPORTING TRUST

Satya Special School's

Genetic Awareness Manual

A Genetic Education & Disability Awareness Initiative

SATYA Special SCHOOL

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Table Of Contents

Genes: The Language of Life | 03

What is Inheritance? | 06

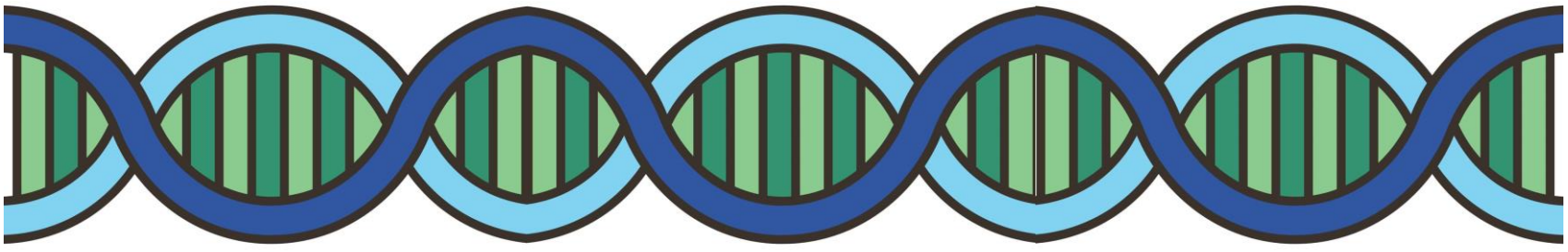
What Causes Genetic Abnormalities? | 09

Disabilities Influenced by Genetics | 11

Risk Factors | 15

Services & Interventions | 18

References | 20

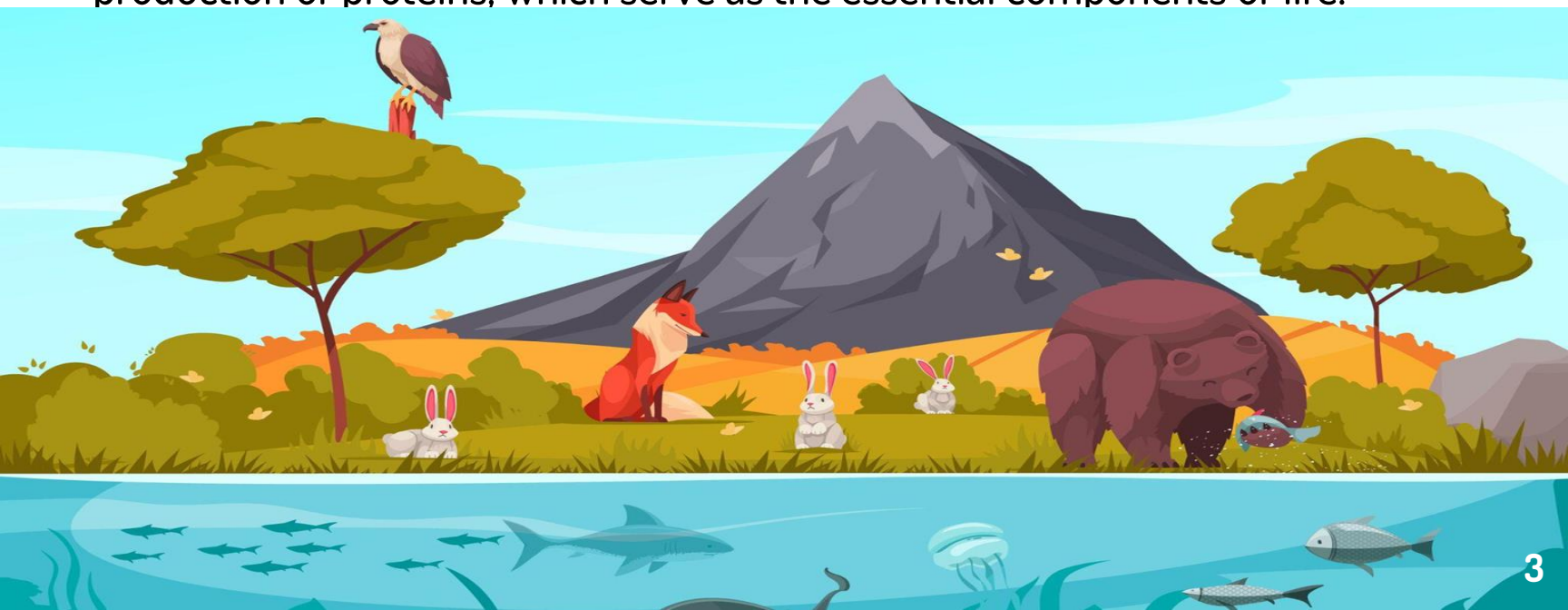


Genes: The Language of Life

We live on a planet with a wide array of diverse life forms. From birds with lightweight bones and marine organisms which lurk at the bottom of the ocean floor, to human beings, Earth has immense biodiversity. The driving force behind this diversity is variations in genetics.

Our genes are the blueprints to life, as they carry the information which determines one's physical and biological traits, features or characteristics. They can be passed down from one generation to the next. It is the differences in genetics which lead to diverse characteristics, behaviors and traits.

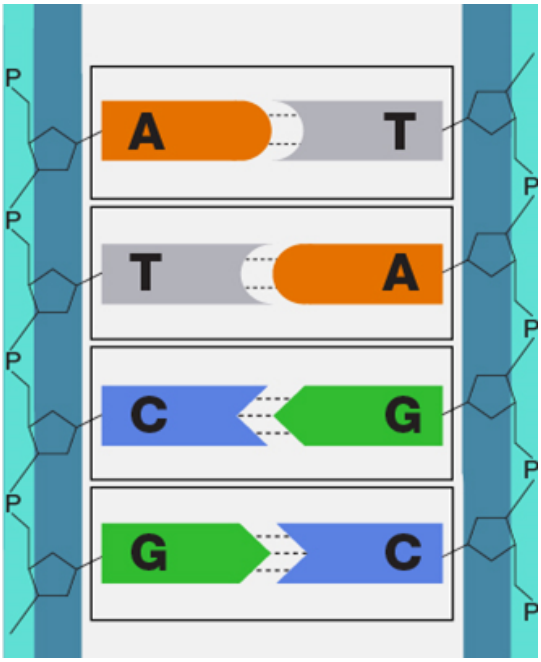
Genes contain instructions which direct your body's functioning and growth. They act as recipes which determine your unique composition. From your eye colour to your height, everything about you is influenced by your genetics. Each cell in the body can carry upto 35,000 genes! Cellular machinery decodes genetic information, leading to the activation of certain genes and the production of proteins, which serve as the essential components of life.



Our genes are composed of a material known as DNA, or deoxyribonucleic acid. The DNA molecule is in the shape of a double helix akin to two twisting ladders. It is made up of building blocks called nucleotides held together through hydrogen bonds.

There are four nucleotides which make up DNA: Cytosine (C), Guanine (G), Adenine (A) and Thymine (T). These nucleotides pair with one another, in the form of a phenomenon known as DNA base pairing. Adenine pairs with Thymine (A-T) , while Cytosine pairs with Guanine (C-G).

These nucleotides are **the language of DNA**. Just like letters, the order of these nucleotides encodes all of the information which determines our body's functioning.



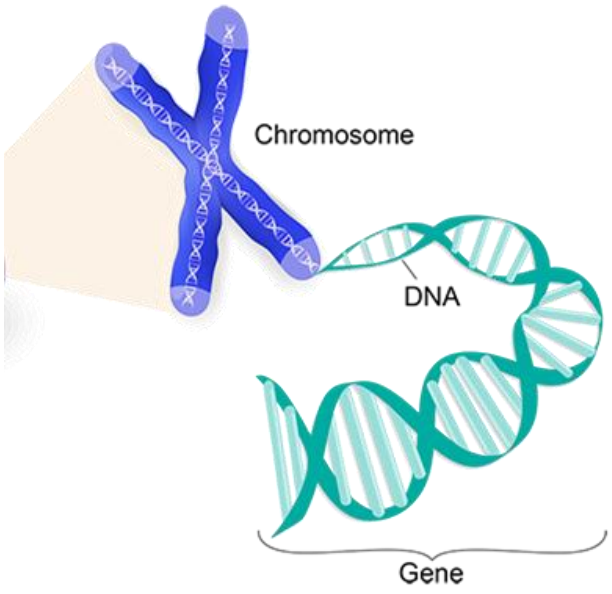
An Analogy:
The nucleotides can be compared to the letters of the alphabet. Just as how letters can be strung together to form words with meaning, the order of the nucleotides determines the biological signal being interpreted. Different sequences of nucleotides lead to the different biological activities.

***For Example**: The nucleotide sequence 'GGC' codes for the production of the protein glycine.*

DNA segments form genes, which can be found in structures called chromosomes. Humans have 46 chromosomes, arranged into 23 pairs of two identical chromosomes each, that are located in the nucleus of our cells. Human beings have:

- 22 pairs of autosomal chromosomes
- 1 pair of sex chromosomes (XY or XX)

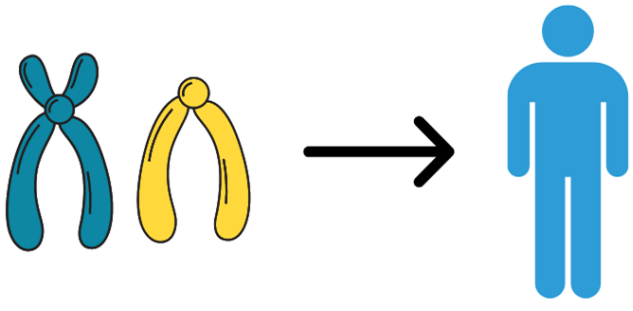
The specific number and arrangement of chromosomes is unique to each species, and varies.

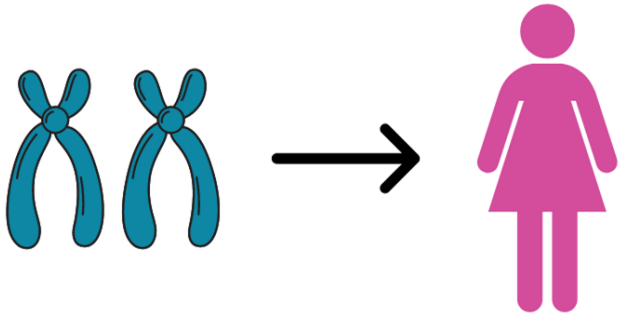


Sex chromosomes determine the **biological sexual development**. In human beings:

Inheriting one X chromosome from the mother, and one Y chromosome from the father results in the birth of a male (XY).

Inheriting one X chromosome from the mother and one X chromosome from the father results in the birth of a female (XX).



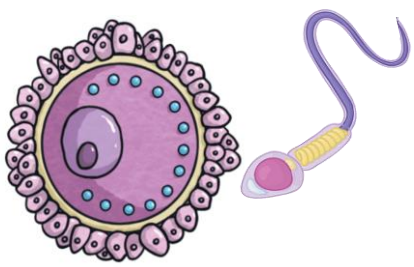


What is Inheritance?

Genetic inheritance is the transfer of genetic information from parents to offspring. A child's genome is made up of a unique combination of genetic traits from **both parents**!

During the process of fertilization, the egg cell of the female combines with the sperm cell of the male to form one single cell known as the zygote. The zygote will develop into the foetus, and contains the genetic information of both the egg and sperm cells.

When fertilization occurs, the sperm and egg each contribute 23 chromosomes, resulting in a zygote with 46 chromosomes, which is the normal number found in human cells. The combination of genetic information from both the sperm and egg results in the creation of a unique genetic identity for the developing child. This means that for every pair of chromosome, one comes from each parent, resulting in children inheriting 50% of their chromosomes from their father and 50% from their mother.



The unique traits expressed by each individual are dependent on the combination of genetic information inherited from the parents.

Certain traits are influenced by a single gene, and are known as monogenic traits. Other traits are determined by a combination of genes and are polygenic in nature. Genetic traits can also be either dominant or recessive in nature. Depending on the interactions which occur between the inherited genes, the individual's characteristics are determined.

Alleles are different versions of the same gene. An individual inherits one allele for a particular trait from each parent. The **combination of alleles** that an individual inherits determines the expression of that trait.

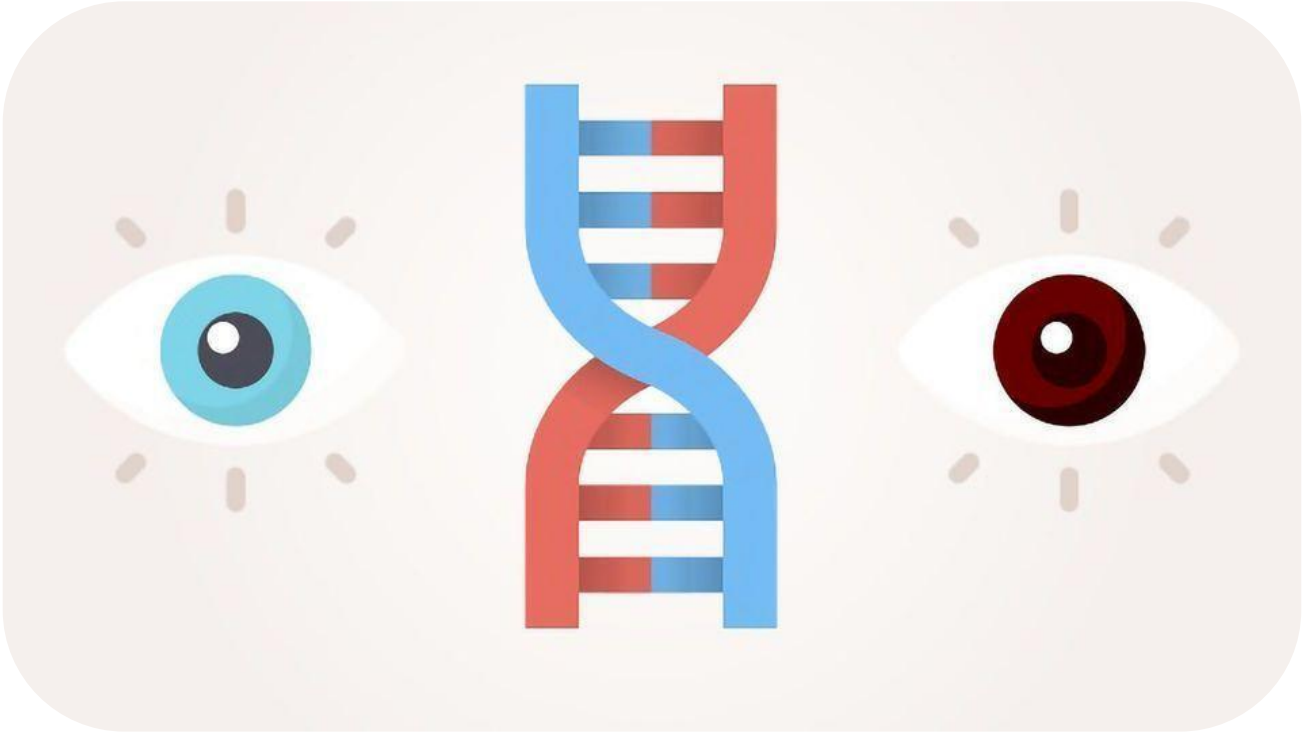
Alleles influence more than just physical traits. They can determine disease risk and predispositions, as well as our responses to drugs, and the environment around us.

For Example:

Case 1: A child inherits the allele for blue eyes from one parent, and the allele for brown eyes from the other parent. As the brown eyes allele is dominant in nature, the child will express **brown eyes** as a trait, even if they also carry a copy of the blue eye allele.

Case 2: A child inherits the allele for blue eyes from one parent, and a second copy of the allele for blue eyes from the other parent. As the blue eyes gene is recessive in nature, it requires two copies of the allele in order to be expressed. The child will express **blue eyes** as a trait in this case.

This example illustrates the significance of genetic interactions and genetic combinations in influencing the unique traits expressed by an individual.

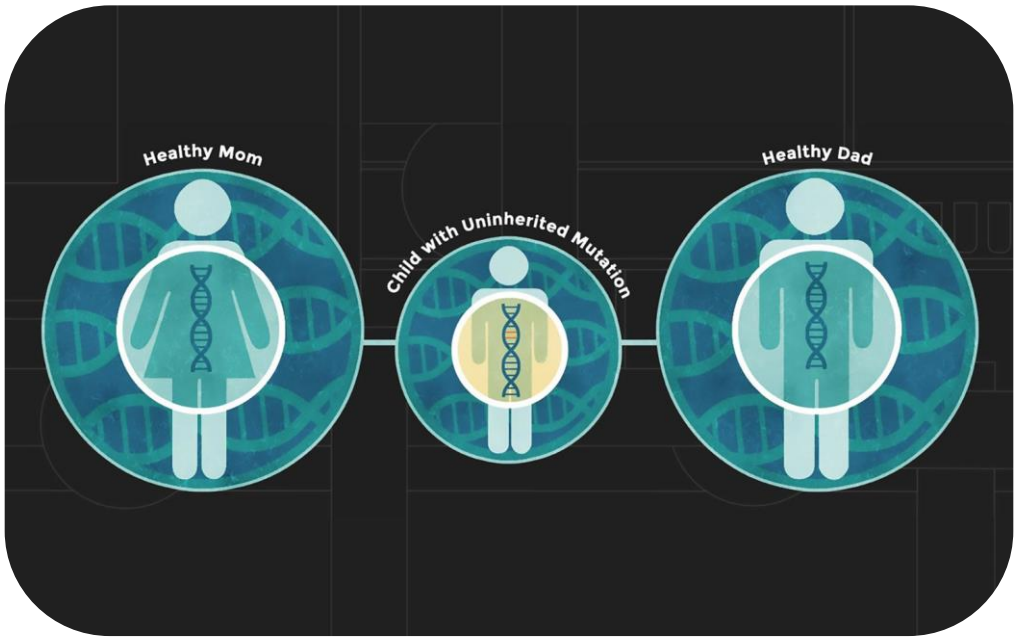


What Causes Genetic Abnormalities?

A genetic abnormality can be defined as as a variation or deviation in an individual's genome, which results in a change in the operation, expression or functioning of their genes. These abnormalities can lead to genetic disorders which can impact the physical or mental well-being of the affected individual. Some common examples of genetic disorders include Down Syndrome, Turner Syndrome, Cystic Fibrosis and more!

In order to further understand these abnormalities, it is important to understand the processes responsible for their occurrence. Genetic abnormalities can be **spontaneous or nonspontaneous in nature.**

Spontaneous genetic abnormalities are unforeseen modifications in the genetic material or chromosome organization. These may occur spontaneously and randomly during cell division or due to errors in DNA replication.



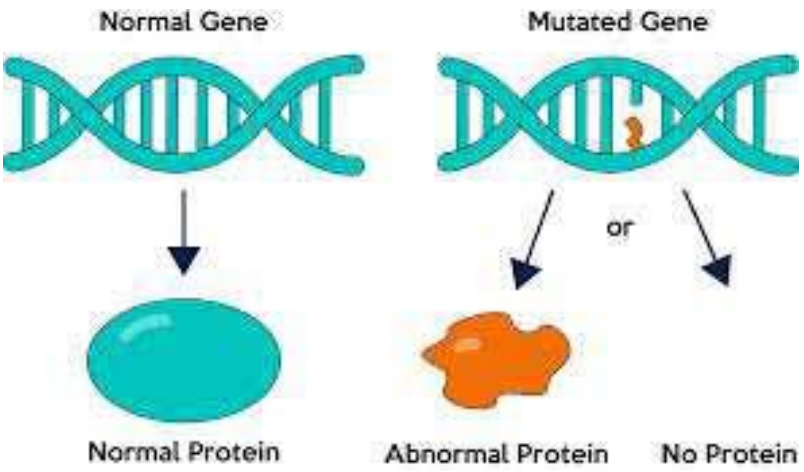
Spontaneous genetic abnormalities include:

1. **De novo mutations**: Mutations which arise spontaneously and are not present in the genetic material inherited from either parent.
2. **Chromosomal abnormalities**: Errors leading to changes in the number or structure of chromosomes.

Non-spontaneous genetic abnormalities are genetic changes that occur as a result of external factors, rather than from spontaneous mutations.

Non spontaneous genetic abnormalities include:

1. **Inherited mutations**: Mutations which are passed down from parent to offspring through genetic material.
2. **Exposure to harmful substance**s: Substances such as chemicals, drugs, radiation, etc can increase the risk of genetic mutations.
3. **Multifactorial inheritance**: A combination of genetic and environmental factors can lead to genetic abnormalities.
4. **Environmental and Social Factors**: Factors such as lifestyle choices, social and biological environments can affect and impact one's genetic composition by affecting gene expression (Epigenetics).



Disabilities Influenced by Genetics

Down Syndrome:

Down Syndrome is a genetic condition which results in significant impact on an individual's health and well-being, causing physical, intellectual, and developmental disabilities. The symptoms of this disorder include cognitive delays, heart defects, and characteristic facial features.

Down Syndrome occurs due to chromosomal abnormality. Most commonly, instead of possessing two copies of the Chromosome 21, individuals possess three copies instead, known as Trisomy 21. The effect of the presence of an extra chromosome varies between individuals. Many people with Down Syndrome lead fulfilling lives, when provided with the appropriate supportive infrastructures.

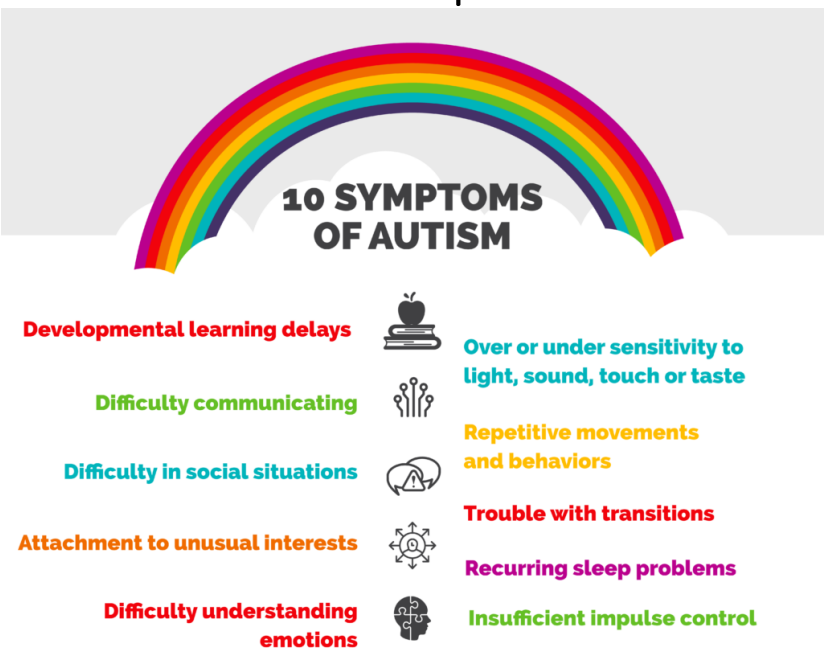
Interventions for individuals with Down syndrome may include:

- Childhood Intervention Programs
- Speech and Communication Therapy
- Special Education
- Medical Treatment,
- Assistive Devices and Technologies
- Support Groups for Inclusion

Autism Spectrum Disorder (ASD):

Autism Spectrum Disorder is a complex neurodevelopmental disorder that has a significant genetic component. It is believed that up to 40-80% of the risk for autism stems from genetic factors. Despite ongoing research, the exact genes involved and how they contribute to autism are yet to be fully understood.

Several genes have been linked to autism, but no one gene has been identified as the sole cause. Rather, it appears that many genes each contribute a small amount to the overall risk. Environmental factors such as maternal health and exposure to toxins during pregnancy also seem to play a



The genetics of autism is an ongoing field of study, and a clearer understanding of the genetic factors involved is expected in the future.

However, it is already evident that genetics plays a crucial role in the development of autism and this knowledge can lead to improved diagnosis, treatments, and interventions for individuals with autism.

Cerebral Palsy:

Cerebral palsy (CP) is a group of neurological conditions that affect movement and coordination. The exact causes of cerebral palsy are not well understood, but it is believed to result from a combination of genetic and environmental factors.

Genetic factors: Some genetic mutations and chromosomal abnormalities have been associated with an increased risk of cerebral palsy. However, the majority of cases of CP are not caused by a single genetic mutation, and instead result from a combination of genetic and environmental factors.

Environmental factors: Some environmental factors that have been linked to the development of cerebral palsy include:

- Premature birth or low birth weight
- Brain damage during pregnancy or birth
- Brain infections (such as meningitis or encephalitis)
- Exposure to certain toxins or drugs

It is important to understand that the genetics of cerebral palsy are complex, and highly vary between individuals. Further research is needed to fully understand the genetic and environmental factors that contribute to the development of cerebral palsy.

Other Intellectual & Learning Disabilities:

A significant proportion of intellectual delays and disabilities are associated with genetics. One such disability is the Fragile X Syndrome. Fragile X syndrome is a genetic disorder resulting in intellectual disability, behavioral issues, and physical difficulties. It is caused by a mutation in the FMR1 gene which prevents the production of the FMR protein, and is one of the most frequently inherited types of intellectual disability.

Diagnosed typically occurs in early childhood and it affects both boys and girls equally. Symptoms may vary from mild to severe, including cognitive and speech impediments, hyperactivity, and social anxiety. Although there is currently no cure, early treatment through therapy, intervention, and special education can assist those affected to manage their symptoms.

Other genetic conditions which cause intellectual impairment and learning difficulties include Williams Syndrome and Turner Syndrome.

Williams Syndrome is caused by a deletion of genetic material from a specific region of chromosome 7. Symptoms include unique facial features, intellectual disability, and developmental delays.

Turner syndrome is a genetic condition that affects only females. It occurs when one of the X chromosomes is missing or partially missing. This results in a total of 45 chromosomes instead of the typical 46 in females. Symptoms include learning difficulties ranging from mild to moderate, ovarian dysfunction, heart defects, a short stature and more.

Risk Factors

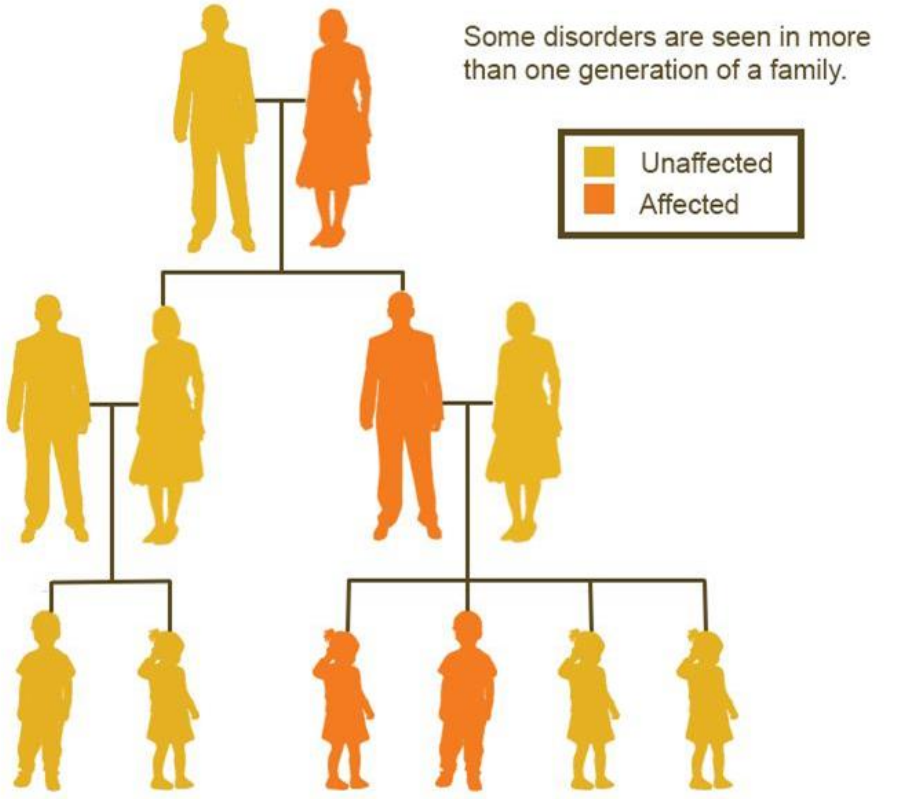
Risk factors are characteristics or conditions that increase the likelihood of developing a particular condition or occurrence. Identifying risk factors is important for several reasons:

1. Early Identification and Intervention: Identifying risk factors can help detect a condition early, when it is easier to treat and before it becomes more serious.
2. Prevention: Understanding risk factors can help individuals take steps to reduce their risk and prevent the development of a disease or health problem.
3. Targeted interventions: By identifying individuals at higher risk, public health providers can tailor interventions to prevent or manage the onset of a condition.
4. Improved Research: Identifying risk factors can lead to a better understanding of the causes of disabilities, which can inform the development of new treatments and management strategies.
5. Improved Quality of Life: Understanding risk factors can allow healthcare providers to improve the overall quality of life for individuals with special needs.

Hence, identifying risk factors is an important part of disability research. By understanding who is at risk and why, healthcare providers and policy makers can take steps to reduce risk and improve health outcomes.

Risk factors for disabilities can vary depending on the type and severity of the disability. Some risk factors include:

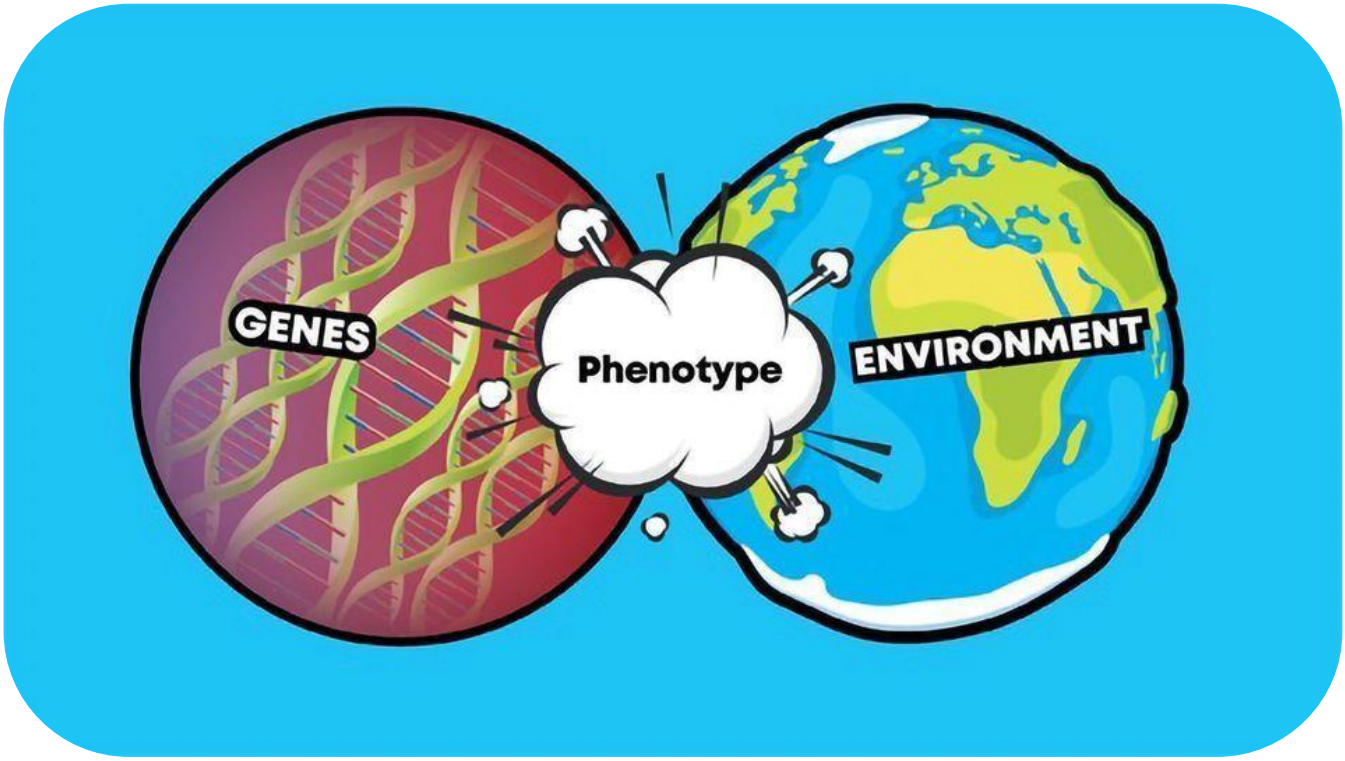
1. Genetics and family history
2. Consanguineous marriages or marriages with a blood relative
3. Premature birth or low birth weight
4. Exposure to infections or viruses during pregnancy or early childhood
5. Chronic health conditions such as diabetes or hypertension
6. Injury or trauma
7. Poor nutrition or lifestyle habits



Environmental risk factors for disabilities can include:

1. Exposure to toxins such as lead, mercury, or pesticides
2. Exposure to harmful substances such as alcohol or drugs during pregnancy
3. Lack of access to proper nutrition or healthcare
4. Living in poverty or socio-economic disadvantage
5. Lack of access to early intervention or educational services
6. Poor housing or living conditions
7. Limited access to transportation or resources

It's important to note that environmental risk factors can interact with genetic and lifestyle factors, and that many people with disabilities do not have a single identifiable risk factor.



Services & Interventions

There are a wide range of services and interventions available for individuals at risk for disabilities, or at risk for having a child with disability. These include:

- Special education programs
- Therapy services (physical, occupational, speech, etc.)
- In-home support services
- Respite care
- Assistive technology
- Support groups for parents and families
- Advocacy organizations
- Accessible recreation and leisure activities
- Transition services for older children entering adulthood.

If you or your child is at risk for a disability, you can also take the following steps:

- Consult a doctor: Get a medical evaluation to determine the cause of your symptoms and the best course of treatment.
- Maintain a healthy lifestyle: Exercise regularly, eat a balanced diet, and get enough sleep to help manage symptoms and improve overall health..
- Genetic Counseling Services: If your family has a history of genetic disorders, an inheritable disability or multiple close relatives with disabilities, genetic counseling services may be a suitable avenue for support.

It's important to take action early and seek support, as early intervention can greatly improve outcomes for those with disabilities.

Genetic Counseling

Genetic counseling is a process that helps individuals and families understand and manage the risks and implications of genetic disorders. It is often used for individuals or families with a history of a genetic condition or for those who are at an increased risk for a genetic disorder.

The process of genetic counseling typically includes:

- Medical and family history review: The counselor will ask about the individual's and family's medical history to determine the risk of a genetic condition.
- Genetic testing: The counselor may recommend genetic testing to confirm a diagnosis or assess the risk of a genetic condition.
- Risk assessment: The counselor will provide information on the likelihood of inheriting or passing on a genetic condition based on the individual's and family's medical history and genetic test results.
- Discussion of options: The counselor will discuss the options available, such as prenatal testing or family planning, to help the individual and family make informed decisions.
- Emotional support: The counselor will provide support and guidance to help the individual and family cope with the psychological and emotional impact of a genetic condition.

Genetic counseling can be an important resource for individuals and families affected by a genetic disorder, helping them make informed decisions and understand the risks and implications of a genetic condition.

References

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Pg 5: https://www.fitnessgenes.com/blog/trait89-telomere-linked-ageing-terc
Pg 7: https://www.youtube.com/watch?v=nDwyHa4nRzs
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