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ORIGINAL ARTICLE

Hereditary Ent Disorders

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ABSTRACT

Background: ENT disorders have a significant impact on a child's development, which is assisted by changes in gene expression.
Aims and objectives: The purpose of this study was to detect and research genetic ENT problems, as well as their forms and therapies.
Methodology: This study was carried out at the Department of ENT Qazi Hussain Ahmad Hospital Nowshera for a period of one year from January 2020 to January 2021. The secondary methodology has been followed in this paper; Different information's has been received that has helped to understand the objectives.
Result: ENT disorder is related with genetic mis-information that is transferred from parents to children.In asia region 2.28 million individuals have reported facing visual impairment and vision loss in 2020. Over 40% of the hearing loss in adult age has also been found to stimulated by genetic disorders that are transferred from parents to children's
Conclusion: this research has illustrated that most of the ENT are curable by surgery and modern approaches.
Keywords: ENT, Cochlear transplant, glaucoma, retinitis

INTRODUCTION

Inherited ENT disorders are malfunctioned conditions or disease in Ears, Throat and Nose. The term inherited means gene associated. The disorders or diseases that are inherited from the previous generation are known as inherited disorders. The major factor that stimulates genetic ENT is congenital defects by birth.

There are several types of genetic ENT disorders such as hearing loss from birth. A study has shown that nearly 60% of infants face hearing loss from birth (Asia Region Healthcare, 2019). Hearing loss at an older age sometimes is not considered as a birth defect, but duringbirth, hearing loss is occurs. There are eye diseases that are also inherited by birth, such as congenital cataracts, retinal degeneration, congenital glaucoma, deformation of eyes, and others. Some of these disorders resulted in complete vision loss from birth. Some of these disorders such as cataract from birth can be removed by laser surgery and can restore the visuals of a child. However, glaucoma from birth is not treatable and leads to complete vision loss. Some of these disorders in eyes such as glaucoma, macular degeneration, are not seen by birth but could occur in adult age. However, both these complications are inherited and genes are responsible for such disease. In the PAK, 2.28 million individuals have reported facing visual impairment and vision loss in 2020. Among them 171 thousandare blind in Britain due to inherited genetic eye disorder (Statista, 2019).

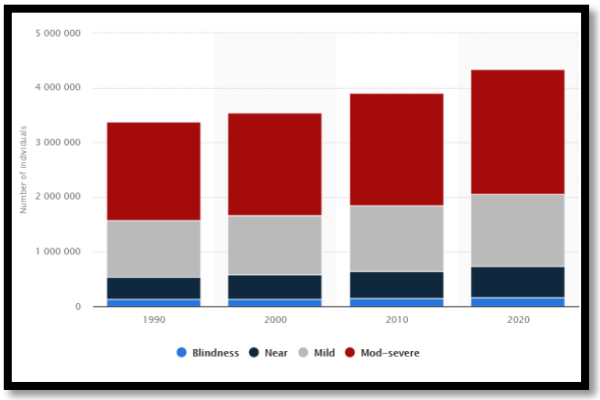

Figure 1: number of individuals having eyes disorders

(Source: influenced by Statista, 2019)

Retinitis pigmentosa is also a degenerative disorder that is night blindness. This is partial blindness. HHT ("Haemorrhagic Hereditary Telangiectasia") disorder is another disease which is associated with bleeding nose disease. This is also known as OWR ("Osler Weber Rendu"), and is a chronic nose bleeding issue and results in vascular dysplasia (Eloy and Musat, 2020). Genetic disorder in formation of joints and bones sometimes led to the malformation of the nose that is also inherited by parental genes. Sometimes joint deformation leads to dysfunction on the nose and in the breathing system. Osteo Disorder which is an inherited disorder affecting the joints and bones also led to the malformation of the nose. Apart from ENT disjunction in lip and chin movement is also considered as an ENT disorder that is also inherited from paternal genes. Geniospasm is such a kind of disorder that led to the dysfunction of lip and chin movement in humans.

Genetic complications inherit from parents to their child. Though sometimes parents may not show any defects and disorders in ENT disease, they pass the defective genes to their children's (Perkins et al. 2019). Intellectual impairment such as AGS ("Aicardi-Goutieres Syndrome"), results from genetic disorders (Sadafi et al. 2019).

The treatment of ENT inherited disorders in recent times has become very popular with gene therapies. Most of the genetic disorders are not treatable such as impairments in hearing, vision loss by birth. high deformation of joints which led to malformation of the nose, which are treatable only by operation. However, a new modern approach and incorporation of technologies has opened up new opportunities in treating such diseases. Gene therapy is also a new approach that has a huge application in the modern medical journey, as well as able to treat retinal disorders which are formed due to defects in genes. However, gene therapies for hearing impairments have not found yet. Though there are cochlear implants that sometimes give the desired results, the chances of complete treatment are minimal. Sensorineural disorder is a neurogenic disorder that affects the hair cells and follicles that are responsible for sensory in ear. However, the treatment for such inherited disorders has not been successful yet. Though hearing aids help to regain a little bit of hearing in individuals.

METHODS AND SOURCES

Secondary method has been followed in this study. This approach has led to collecting different results from different sources.

Multiple websites have been used to collect data regarding ENT disorder. Both qualitative and quantitative data is used in this study. Data are also collected from previous journals and books that are available on “Google Scholar". As it is a reliable website and also contains a large number of journals and articles that have fulfilled the information that is needed. Articles regarding genetic disorders have helped to understand the reason and factors such as haemorrhagic factors that are one of the major reasons in nose disorder and nose bleeding. This disorder is also inherited by genetic defects caused by gene mutation. According to a study there is a multifactorial disorder that is also inherited by genetic disorder (Zelenkova et al. 2019).

Majority of data regarding Hereditary ENT disorders has been collected from the Department of ENT Qazi Hussain Ahmad Hospital Nowshera based databases. It is a professional body that is developing excellence in ENT. The professional body of ENT also represents the ENT surgery to provide better treatment for the ENT disorder patients. This website has also helped to achieve information from ENT events in this study. This site has also helped to collect information regarding ENT vitalities in pandemic situations due to covid 19.

Another database from DHQ Hospital nowshera Pakistan, has helped to gain information on ENT surgery and treatment. The ENT disorder that may develop with age or inherited from genetic disorders and their treatment has been discussed in this website. This site has also illustrated different principles of ENT surgery that are essential in ENT treatment. There are six types of ENT surgery principles that have been discussed in the above-mentioned site (RCSE PAK, 2019).

The NHS (“National Health Services”) PAK has also taken as a source of data collection in this study. This site has helped to gain information about the ENT disorder and their treatment in the PAK. This site has helped to understand the otorhinolaryngology which is the surgery of ENT to fix the ENT disorders. This site has shared the insights of the ENT surgery and how they work. The interventions and studies on ENT have been discussed in this site also. The different conditions of ears and their surgery has been found on this site. This particular site has helped to understand the different conditions of ear disorder and how different surgeries helped to treat them (NHS PAK, 2019). Some information from, ENT audiology News has helped to understand the association of genetic neurological disorder and ENT disorder in children. This site has also helped with information with several neurological and other disorders and how they affect the ENT (ENT audiology News, 2019).

An PAK alliance site of ENT has helped to understand the support and treatment of rare ENT disorders that are inherited from parents to children. This site is an open forum that is supporting people from different backgrounds that suffer from ENT . This site also gives information about the charity and funds that helped patients suffering from ENT to receive treatments and surgeries. There are many health care’scentres and hospitals that voluntarily helped ENT surgeries in children

The different sources in the secondary method have helped to achieve both qualitative quantitative data to enrich the study. The publications from Parliament, they have also been taken as a data source to understand the government perspective on ENT disorder and their treatment. This site from the pak government has also discussed the funds and donations from the government to ENT studies and research purposes (Parliament, 2019).

RESULTS

Multiple data from different websites have helped to understand the factors that stimulate genetic disorders and the rate of genetic disease among the Pakistani population. According to WHO in the future more people will face hearing disorders and the number will be 2.5 billion (WHO, 2019)

Table 1: different disorder and their treatment

Hereditary ENT disorders	Causes	Effect	Treatment
Hearing loss by birth	Intrauterine infection during pregnancy, Viral infection, bacterial infection that passes through genes from parents to children. Sometime Asphyxia during birth also led to ear disorder	Hearing loss, Partial hearing loss, Loss of ear tissue are the effects of ear disorder. Dysfunction in haring joints also causes hearing impairments	Hearing aids are the major treatment. Cochlear transplant and Joints replacement in hearing joints also help in treatment
Eyes disorders and vision loss	Mutation in gene and Monogenic disorder during pregnancy is the major cause that leads to child vision disorder. Change in chromosomal structure	Development of child glaucoma, Retinitis pigmentosa are the major eye disorders.	Some surgeries may help to treating the retinitis and cataract, though there is no treatment for glaucoma
Nose disorder	Nose dois orders are mostly inherited in joints and bone disorder that leads to dysfunction and deformation of nose	Difficulties in breathing, nose bleeding are the nose disorder that are inherited to children from parents	Nose joints surgeries helped in fixing the joints

There are different types of ENTdisorders that have become treatable with new approaches. However, the British government has focused on development of the individuals that are suffering from such disorders. “The British Deaf Association” has revealed that more than 3 children among hundred suffer from hearing impairments by birth (British Deaf association, 2019). However the prevalence of hearing loss according to age in the PAK is also very high, over 40% . the hearing loss in adult age may also be stimulated by genetic disorders that are transferred from parents to children’s (Statista PAK, 2019)

(Source: influenced by Statista , 2019)

In order to enhance the treatment and to cure such ENT disorders, the PAK government has focused on developing new treatment centres based on modern treatment approaches.

(Source: influenced by PAK Parliament, 2019)

The inherited eye diseases are more severe and also there is a lack of treatments of eye disease such as IRDS (“Inherited Retinal disorders' '). The IRDS are generally referred to as a heterogeneous disease that has been passed through the parental genes to their children and the progressive nature of this disorder resulted in degradation of photoreceptors of epithelial cells in human eyes. This disease has a wide occurrence around 2 million people worldwide. However, the neurological syndrome may stimulate this disease in most cases. This particular disorder also has syndromic occurrences such as “Senior-loken” syndrome, “Bardet Biedl” syndrome, and “Usher syndrome”. According to a study more than 208 genes are responsible for the development of IRDS (Wang et al. 2018). However, “Non-generation-sequencing” is the new approach that has been applied in treatment of IRDS.

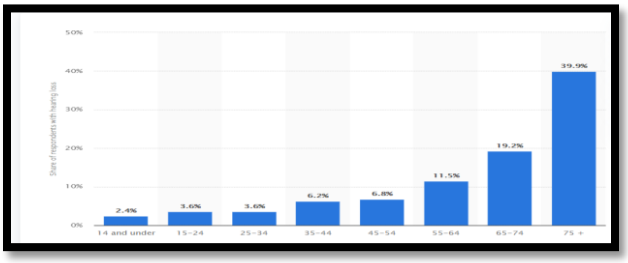

Figure 2: number of individuals of hearing loss in this study

Type	Principal Investigator	Organisation	Title	Amount Awarded*
Grant	Professor J Connell/ Professor M Caulfield	University of Glasgow/ Queen Mary, University of London	The MRC British Genetics of Hypertension Study	£5.1 million
Grant	Professor T Moffitt	Institute of Psychiatry	Gene—environment interplay in early-onset psychopathology	£5.4 million
Grant	Professor J Golding	University of Bristol	ALSPAC: A reference population for genetic and environmental epidemiology	£8.3 million

Figure 3: different organization and spent amount on ENT treatment by British government

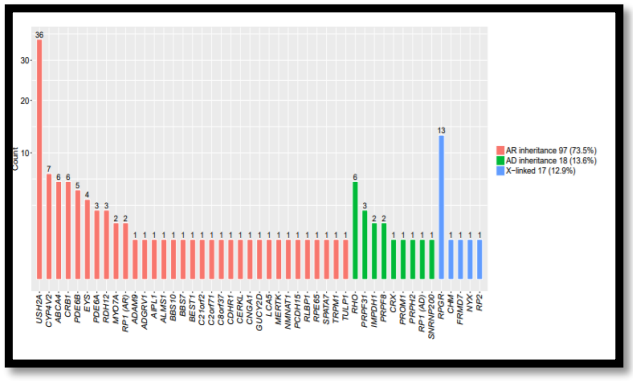

Figure 4: inherited genes in IRDS

(Source: influenced by Wang et al. 2018)

The inherited IRAS are one of the major causes that influence vision loss from birth. Gene variability and their expression patterns are also responsible for the inheritance in eye disorder, which is sometimes expressed in adulthood and may also present from birth.

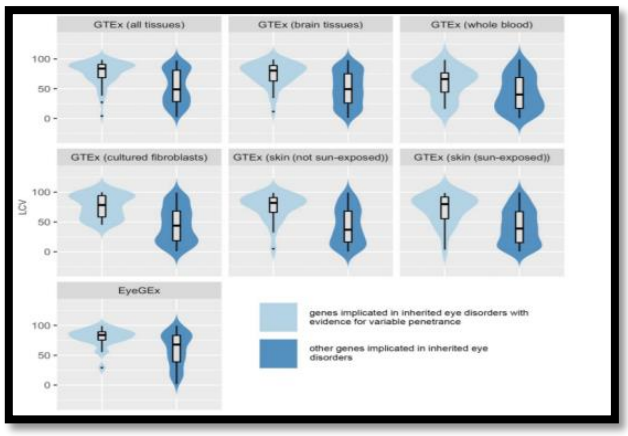

Figure 5: gene expression pattern and eye disorder

(Source: influenced by Green et al 2020)

The variability in gene expression that aligned differently and this different expression sometimes transferred to the children's gene from the parents, which ultimately led to eye disorder (Green et al 2020). Such inherited nose disorder mostly resulted from muscle deformation. The congenital diaphragmatic hernia is the diaphragm formation disorder that is developed in children. This led to the complex and difficult breathing in children at very early age (Chandrasekharan et al. 2017)

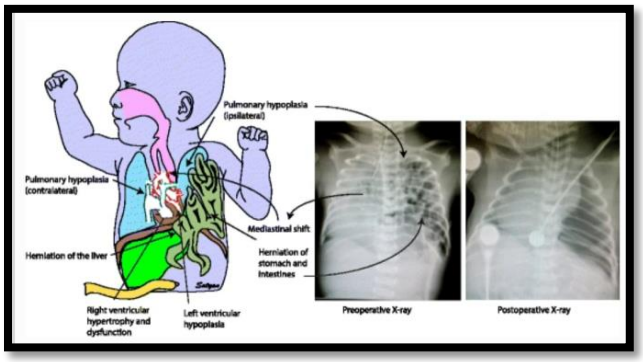

Figure 6: Diaphragmatic hernia in children

(Source: influenced by Chandrasekharan et al. 2017)

DISCUSSION

The genetic disorder in ENT in children and adults has a huge impact on their life. This congenital disorder not only affects physical health but also affects mental health. The different disorders such as malformation in nose, dysfunction of nose joints, hearing loss and others also have social implications on an individual. Such genetic disorder also influences the risk of survival due to different ENT disorders. There are several diseases that have been found in eye disorders which influence blindness and loss of vision in children. However, many disorders related to ENT do not show any symptoms from birth but develop with age when additional symptoms of different disorders occur with growing age (Wang et al. 2018). There are associations of deformation with any disorder which enhance the severity of a disease such as pragmatic hernia associated with breathing shortness and influence it. There are cases that have shown individuals that having neurological disorders such as down syndromes, Parkinson disease or others has the chances to grow ENT disorders as well. Such disorders hamper speech, or sometime associated with breathing shortness or hearing loss. Sometimes these genetic disorders related to ENT are not cured with treatment and have impacted the social life of a patient. Deformation from birth also led to the slow development of a child. The different ENT disorders have a huge mental impact on a child and may also lead to anxiety or depression. Genetic disorder are the effect of a missing gene sequencing or multiple variations during gene expression. These disorders or disease may not present in parents but sometimes the misinformation is transferred to the children that cause multiple genetic disorders such as ENT disorders. Sometime when a parent is suffering from disorders such as blindness, IRDS, partial blindness and others, the chances of passing the genetic information to the next generation genes may resulted in developing genetic disorders to the children (Wang et al. 2018).

There are other factors rather than genetic misinformation that also causes genetic disorder and may not inherited from the parents but developed during birth. Infection that may be caused due to bacterial or viral invasion, also found to influenced different and variable missing gene expression and resulted in ENT disorders. However, these events rarely occur. Asphyxia is such a condition that promotes learning loss during birth due to lack of oxygen and may not be curable in adulthood. However, the modern approaches in treatment such as high technology-based surgeries have helped to cure completely or partially these disorders. The enhancement of such disorders in human beings

has been increasing in recent times. The above study has shown that in the PAK, the number of hearing impairment and blind individuals are increasing which does not support the development of the whole community in a country. The increasing rate also indicates that lack of good treatment approaches and opportunities. However, the PAK government has invested in many organizations and authorities that are involved in research and development of new treatment of such disease and disorder. The expense is the major barrier that limits individuals from poor backgrounds to receive treatments. However, many authorities in the PAK are voluntarily working on providing treatments to cure such disorders. However, the NHS PAK has majorly focused on collaboration of the PAK government and the health authorities to investigate the necessity of treatment and do the essentials (Genetic Alliance PAK, 2019).

CONCLUSION

Overall, it can be concluded that disorders which are hereditary ENT, are one of the major problems in contemporary time.These are majorly found in people with different ages, worldwide. This study has helped to conclude that there are different types of ENT disorders that developed due to defects in gene expression. It can be also concluded that some modern treatment approaches such as gene therapy, gene expression variations help to treat such disorders. This study has also helped to conclude that new approaches and collaboration of different health services would be able to treat such disorders. ENT infections has become a severe problem that also limits the development of a child. Various studies and data in this study has helped to conclude that some genetic disorders related to ENT are not curable. However, transplant of organs and issues such as cochlear transplant, eye tissue transplant may help to cure such disease.

However, there are few recommendations that will help to understand the disorder and also help to identify the right treatment. The government collaboration and activities will help to raise funds so that everyone can receive treatments according to their needs. Awareness programs are also very useful so that people understand the reason and the cure of such disorders. Sometimes people have lack of access to prevention and cure, hence awareness programs will help them to understand the actual reasons of such disease and how to cooperate with them. Overall social awareness and compassion towards the sufferers are needed so that they will also feel important like the normal individual and it will also help them to develop confidence. Children who are suffering from ENT disorder sometimes do not get the opportunity of learning and studying, hence the Government should focus on developing special schools and listening facilities so that they can also receive the same education as others.

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