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Human Genetics

GENETIC DISEASES, COUNSELING AND REHABILITATION IN DENTISTRY: A NARRATIVE VIEW AND SYSTEMATIC APPROACH

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ABSTRACT **Introduction:** The oral cavity contains a plenitude of the signs of diseases in the human body. Some of these dysfunctions are influenced by the genetic arrangement and are considered as hereditary. The patient visiting a dentist is most of the time concerned about the addressal to their present chief complaint only. However, the onus lies on the dental surgeon to recognize such abnormalities and the future challenges which can be posed to the concerned patients. **Aim:** Identification, management, counseling of patient suffering from genetic syndromes on examination of oral cavity. **Methods:** The step-by-step methodical approach involving careful history, specific investigation protocol, pedigree analysis and genetic testing to be followed in the management influenced by sufficient knowledge on the subject and the sources that can be utilized are being discussed and summarized here. Also, discussion of appropriate sources for knowledge, organizations which can be tapped as a part of genetic counseling. **Conclusion:** Genetic counseling involves catering to emotional, psychological, medical, social, and economic concerns of patients and their kins. Increasing awareness about current and potential future health issues for these patients as well as their next generations is crucial.

KEYWORDS : Dental diseases, Genetic literacy, Genetic counseling, Management

INTRODUCTION

Genetic variations have always been a part of the evolutionary world. While combination of 'good' genes is known to cause revision in phenotypic and neurological characteristics in humans. Unfortunate deletions, mutations or recombination cause implications which have to be correctly understood and managed by a medical or dental professional.

Genetics effects the aggregate part of human body. Genes are basic constituents of each cell and govern all functions and well-being of individual. Genetic variations are cause of many inherited diseases. These maybe single or polygenic involving multiple number of genes causing severe craniofacial abnormalities.

Dental surgeons have a responsibility to identify these deviations because of their prodromal effects and signs in oral cavity. Also, educating dental patients and their kins brings a huge responsibility which implies having a sound knowledge, training and proper referrals.

GENETIC LITERACY

The Human Genome Project (HGP) began in 1990 and concluded in 2003, being the largest collaborative biological project ever undertaken. The Human Genome Project was an international scientific initiative by the National Human Genome Research Institute (NHGRI) aimed at determining the base pairs that compose human DNA and at identifying, mapping, and sequencing all the genes in the human genome from both physical and functional perspectives. The HGP was declared complete on April 14, 2003, covering approximately 92% of the genome. The level of a "complete genome" was achieved in May 2021, with only about 0.3% of bases remaining due to potential issues. The final complete assembly was completed in 2022.

The Human Genome Project's most notable achievement, producing the first complete sequence of the human genome has provided essential insights into the human genetic blueprint. This accomplishment has greatly accelerated the study of human biology and enhanced medical practice.[1]The benefits of discovering sequence of all the 23 pair of chromosomes is to be able to identify by testing any deviations found from the normal.

Certain sites are available where the characteristics and traits in a patient can be compared with the list of diseases/deformities catalogued. Important online resources for information about genetic conditions include **GeneReviews** (www.ncbi.nlm.nih.gov/books/NBK1116), **MedGen** (www.ncbi.nlm.nih.gov/medgen), and **Online Mendelian Inheritance in Man** (OMIM; www.omim.org/). OMIM can also be used to find lists of internal and external clinical features associated with specific genetic conditions.[2]

Other sources include **Genetic and Rare Disease Information Centre** (GARD) site where Information Specialists offer individualized help to connect with concerned information.

National Center for Advancing Translational Sciences (NCATS) a part of NIH provides free materials and resources designed to assist patients, caregivers, patient support organizations, healthcare providers, and scientists in understanding rare diseases and promoting research advancements.

The **Undiagnosed Diseases Network** (UDN) is an NIH-funded research study dedicated to enhancing the diagnosis and care of patients with undiagnosed diseases.[3]

National Organization of Rare Disorders(NORD) helps to understand rare diseases through its database and to ensure that people living with rare diseases have access to the same resources as any other population.[4]

GENETIC COUNSELING

Genetic counseling is **defined** as the process of assisting individuals in comprehending and adjusting to the medical, psychological, and familial implications of genetic factors contributing to disease. It encompasses first analyzing family and medical histories to evaluate the likelihood of disease occurrence or recurrence.

Then providing education on inheritance patterns, testing options, management strategies, prevention, available resources, and current research.

Finally, offering counseling to encourage informed decision-making and adaptation to the risk or condition.

GENETIC COUNSELING IN DENTISTRY

It is essential for dentists to be prepared for identification and management of various genetic diseases and disorders. Pediatric dentists are often the first healthcare providers to identify dysmorphic features in a child.[5] Also, dental professionals have a unique opportunity to monitor the growth and development of preadolescent and adolescent patients during critical periods of change.[6]

Fig.1. Steps in Genetic Counseling

Steps for a smooth approach that need to be taken are as follows-

1. Adequate knowledge/education

- Courses related to diagnosis, handling of patients with genetic diseases can be incorporated in the dental program.
- Continued dental education by the government dental body for updating knowledge on genetic abnormalities affecting the craniofacial/oral region.
- On ground training. Budding dentists can be sent to various genetic clinics around the country during internship.

2. Identification Of Patient

During professional practice, application of this knowledge is required the minute such patient enters the clinic for some dental advise or treatment. It is critical to be able to identify such potential patients.

Craniofacial and oral abnormalities are often identified by a dental surgeon. Tooth structure abnormalities with enamel or dentin deficiency, oligodontia, microdontia or delayed eruption and shedding is known to be related with a couple of systemic deformities and syndromes.

Few relatively rare genetic cases affecting orofacial growth and development such as-

CHARGE syndrome is a rare genetic disorder characterized by the presence of coloboma, choanal atresia or stenosis, cranial nerve dysfunction or anomalies, and distinctive features of the external, middle, or inner ear. The condition can severely impact overall health and may be further complicated by cognitive or intellectual impairments, feeding difficulties, and behavioral challenges. The patient is mostly unable to suck or swallow, and has micrognathia.[7] Oral hygiene maintenance and fluoride treatments are required in such cases.

Pycnodysostosis, an autosomal recessive disorder is characterized by short-limbed short stature and a distinct facial appearance including a convex nasal ridge and a small jaw with an obtuse mandibular angle. There are variants in CTSK genes found through molecular genetic testing. Treatment includes craniofacial and neurosurgical care for conditions like cleft palate, craniosynostosis, and maxillary and mandibular hypoplasia. Management also involves pulmonology and sleep medicine specialists for obstructive sleep apnea, and dental and orthodontic care for dental anomalies.

3. After presumptive diagnosis, an informed consent from the patient/kin is taken for further identification.

Two major steps followed for confirmation are mainly **genetic testing** and **pedigree construction.**

A. Pedigree

Creating a pedigree is a concise way to categorize family information, effectively summarize the data for the analysis of inherited traits. The pedigree tree also helps to identify if the mode of inheritance of disease is dominant or recessive.[8]

Generally, last three generations of patient with paternal and maternal relatives upto third cousins, uncles and aunts are involved.

B. Genetic testing

Genetic screening refers to tests conducted on a population-wide basis to identify individuals at risk for certain conditions. Genetic testing, on the other hand, involves specific assays used for definitive diagnosis, which may be performed due to positive screening results, family history, ethnicity, physical markers, or other reasons.

Different Types of Screening:

a. **Diagnostic Testing:** Used to diagnose or rule out a specific genetic or chromosomal condition.
b. **Carrier Testing**: Identifies individuals who carry one copy of a genetic mutation that can cause a genetic disorder when present in two copies.
c. **Predictive and Presymptomatic Testing**: Identifies gene mutations associated with disorders that may occur after birth or later in life.[9]

4. Management

After results confirmation are received, proper guidance should be provided. It is seen that patient families are often confused and are in a state of shock after the revelation of such knowledge. They should be dealt with empathy and patience.

Effective communication is required for genetic counseling. Counselors have to be good listeners who are sensitive about their patients test results**.** Facts should be presented in an easy-to-understand manner to simplify a complex information. [10]

Genetic Counseling can have Prospective and Retrospective Approaches

Prospective Genetic Counseling: This approach focuses on the true prevention of genetic diseases. It involves identifying individuals who are heterozygous carriers of a specific genetic defect through the screening procedures. These individuals are then informed about the risks of having affected children if they marry another carrier of the same gene.

Retrospective Genetic Counseling: Currently, most genetic counseling is retrospective, occurring after a hereditary disorder has already manifested within a family. [11]

Examination of such patients may be done with utmost patience.
Some modifications in their management maybe required.

- Prophylactic care is advised to avoid extensive treatments in future.
- Hand scaling maybe performed if ultrasonic scaling is not comfortable to the patient.
- Fluoride application of teeth is done to avoid caries.
- In many patients with poorly developed maxilla, mandible, orthodontic treatment maybe proposed along with fixed or removable appliances.
- Use of electric toothbrushes, water pick system is advised if manual dexterity exists.
- Brochures and information booklets pertaining to special oral care maybe handed over.
- Sharing of sources, organizations with patient's family can have continued support.
- Counseling aids can also be developed by using PowerPoint slides to present text and diagrams for educating families. Conducting an internet search, particularly on websites of national organizations or support groups related to genetic condition, can help find relevant content, photos, and diagrams to include**.**

Referrals

A team of psychiatrist, physicians, neurologist and specialists should be roped in for guidance and other treatments. Sometimes, physiotherapist, speech therapists are also required for such patients. Overall motive should be to improve or at least manage existing health condition of patient.

Awareness

An important aspect also involves awareness of possible effect of such genes in future generations. All along, maintaining confidentiality and privacy of the patient.[12]

DISCUSSION

" Genetics is an exploration, from unravelling our ancestry to finding cures for diseases." **Robert Waterston.**

Oral diseases result from complex aetiology and influence, with genetic factors playing a significant role in their development and resolution.

It has been assumed that relationships are fundamental to genetic counseling. The strength and quality of the relationship between the genetic counselor and the patient are just as crucial as the genetic information provided.[13]

Important **Roles** (denoted as crucial '**R**'s) related to genetic counseling of a dental patient as summarized would be-

Table 1

i . Reduce: Increase awareness among patients and their families about genetic diseases to minimize health damage.
ii. Reform: Adapt treatment methods as needed based on the patient's condition. For instance, a compliant patient can receive normal chairside treatment, whereas a non-compliant patient may require management under sedation.
iii. **R**estore:

-Speech: Use occlusal therapy or palatal appliances for perforations or developmental defects.
- Chewing: Treat carious teeth with restorations, root canal treatments, or extractions.
- Breathing: Remove mouth obstructions and address open bites and facial deformities.
iv. **R**ecover: Although permanent cures may not be possible, reducing morbidity is crucial.
v. **R**eliance: Develop rapport with patient and family by providing continuous support and availability at all hours of need.
vi. **R**emember: Instill hope and work with certitude for the benefit of such patients.

Some brilliant advances in genetic field like molecular biology, gene therapy and artificial intelligence also give a hope of changing course of management in future.

Molecular biology has always promised to transform medicine from a stroke of luck into a rational pursuit grounded in understanding the mechanisms of life. Genomics will expedite the integration of molecular biology into medical practice.

Artificial Intelligence (AI) is revolutionizing clinical genetics. AI algorithms can swiftly and accurately analyse large volumes of genetic data, resulting in more precise diagnoses and personalized treatment plans.

Advancements in genetic sequencing, the human genome project, and medical genetics have uncovered new disease-causing genes, improved diagnostic accuracy, and provided new drug development targets. Emerging technologies like **single-cell genome sequencing**[14,15], **CRISPR-Cas9**[16], and **liquid biopsies**[17] are poised to further shape the future of clinical genetics, offering more precise diagnoses, personalized treatments, and enhanced disease prevention strategies.

CONCLUSION

Genetic counseling conveys crucial information to address the concerns of individuals or families, helping them make informed medical decisions. Counselors assist clients in coping with and adapting to the emotional, psychological, medical, social, and economic implications of their test results.[18,19]

Also, as these technologies and processes continue to evolve and become more widely adopted, the future of clinical genetics appears promising.

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