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Editorial

Cytogenetics: A New Frontier in Understanding Genetic Disorders

Waleed Hassan[1*], Ghazala Qamar[2]

[1]Assistant Editor, Journal of Islamabad Medical & Dental College, Islamabad, Pakistan

[2]Consultant Haematologist, Chughtai Institute of Pathology, Lahore, Pakistan

[*]Correspondence: waleed.hassan@imdcollege.edu.pk

Since the complex etiology of genetic disorders are sometimes masked within the microscopic observation of chromosomes and therefore pose a challenge to modern medicine. Knowledge of human health and disease can be revolutionized by using 'cytogenetics', which can serve as a potent lens to solve these genetic dilemmas. Cytogenetics, the field of genetics that studies chromosomes and their role in inheritance, is becoming increasingly crucial in the diagnosis and therapy of genetic disorders and cancers. In Pakistan, the field has gained attraction as healthcare practitioners understand its usefulness in tackling the increased incidence of hematological malignancies and genetic disorders. It has transformed medical diagnosis by offering previously unknown insights into chromosomal abnormalities. It provides a crucial judgement into the genetic foundations of various chromosomal disorders, cancer research to prenatal screening.[1]

Cytogenetics is the study of chromosomes, which are substantial protein and DNA strands that make up the majority of a cell's genetic material. One pair of sex chromosomes plus 23 pairs of autosomal chromosomes (46, XX in females and 46, XY in men) make up the human genome. Chromosome alterations may be a sign of an underlying hereditary condition, or malignancy which is phenotypically expressed as a disorder. A detailed investigation, treatment strategy, or evaluation of the efficacy of a treatment plan can all be aided by cytogenetics. Most living cells' nucleus have a thread-like structure of proteins and nucleic acids that carry genes, which collectively make up the genetic material.[2]

Variance in the chromosome might be structural or numerical. Anything less than a full set of 46 chromosomes indicates a change in the amount of genetic material present and may lead to retarded growth and other abnormalities. The chromosome that is changed determines the importance of the issues and how serious they are in the case of structural modifications. The type and severity of disorder can vary based on individual to individual, even if the same genetic abnormality is present. Chromosomal karyotyping examines a person's chromosomes to determine if the right number is present and to determine if each chromosome appears normal.[3]

In terms of genomics, deletion is a sort of mutation that happens when one or more nucleotides are lost from a section of DNA. Any number of nucleotides can be lost during a deletion, ranging from one nucleotide to the loss of an entire section of a chromosome. One or more nucleotides from the DNA segment are lost in this particular sort of frameshift mutation. DiGeorge syndrome is an illustration of deletion mutation. A little portion of chromosome 22 is deleted, leading to the condition known as DiGeorge syndrome.[4] When a segment of one chromosome is translocated and inserted into the interstitial space of another non-homologous chromosome, the process is known as chromosomal insertion. Alternatively, intrachromosomal insertion happens when the segment is placed into a different location of the same chromosome. [5]

Depending on the type of chromosomal abnormality, there may be a variety of signs and symptoms, such as an abnormally shaped head, cleft lip (openings in the lip or mouth), low birth weight, mental and physical disabilities, heart, intestine, kidney, and stomach defects, reduced muscle mass, ambiguous genitalia, and distinctive facial features (widely spaced eyes, small and low ears, flattened facial profile. Early intervention for patients and their families through genetic counselling and assistance with care planning is crucial for improving their

quality of life. By employing various mitogens, which improve the ability of mitotic stimulation and provide good mitotic indexes that aid in the interpretation of the results, karyotyping can be used to diagnose the suspected chromosomal problem.[6]

To identify chromosome abnormalities such as aneuploidy and structural abnormalities, chromosomes are examined during cytogenetic testing. 23 pairs of chromosomes make up a typical human cell, containing 22 pairs of autosomes and one pair of XX or XY chromosomes for sex. Aneuploidy is characterized by the presence of one or more additional chromosomes (such as 47 XX +21 or 48 XXXY) or the absence of chromosomes (such as 45 XO). The most prevalent aneuploidies include Turner syndrome (monosomy X), Down syndrome (trisomy 21), and Edward's syndrome (trisomy 18).[7]

Although these developments, a number of obstacles prevent cytogenetics in Pakistan from reaching its full potential. The poor understanding of patients and healthcare professionals regarding genetic illnesses is a significant barrier. resulting in delayed diagnoses and lost chances for early intervention. The problem is further complicated by the fact that genetic screening focusing on public health initiatives is still in infancy.[2]

Human resource capacity in cytogenetics is another critical area needing attention. Many laboratories still rely on traditional methods that may not suffice for complex cases. There is a pressing need for specialized training programs to equip healthcare professionals with the necessary skills to perform advanced cytogenetic analyses accurately. Nevertheless, cytogenetics has far more role and potential than simple diagnostics. It acts as a link between personalized therapy and genetic knowledge. Researchers can potentially lessen the effects of genetic disorders by developing targeted therapies through identification of specific chromosomal abnormalities.[9] In comparison to conventional generalized medical treatments, this strategy marks a fundamental change, i.e. Cytogenetics has a bright future. New technologies keep improving our capacity to identify and understand genetic variants with expanding accuracy. We are heading closer to a time when genetic disorders will be more accurately diagnosed and may be cured as well.

Advances in technology are broadening the scope of cytogenetic studies. Researchers may now examine genetic material more precisely, Thanks to the Next-generation sequencing tools. This includes discovering previously undetectable chromosomal variants, which may reveal genetic susceptibilities to illnesses long before symptoms appear.[10] Think about the moral situation with genetic screening. Early detection of possible genetic abnormalities has significant medical benefits, but it also raises concerns about patient confidentiality, privacy, and the psychological effects of genetic knowledge. For example, prenatal screening offers difficult choice for parents regarding genetic variants that could affect the life of their future offspring.[11]

Future prospects for cytogenetics in Pakistan appear bright, but support from a range of stakeholders is needed. Early identification of at-risk people can be facilitated by extending public health initiatives to include comprehensive genetic screening programs. Proactive steps like this could greatly lower the prevalence of genetic diseases and enhance public trust.[12] It is also crucial to incorporate cytogenetic services into basic healthcare systems. Clinical labs and academic institutions working together can enhance diagnostic capabilities and promote innovation.

In addition to improving treatment procedures, this would add significant knowledge about genetic diseases to the global database of knowledge.
Additionally, it is critical to educate patients and healthcare professionals on the significance of cytogenetics. Campaigns for education can empower people to get genetic testing when needed and motivate medical providers to include these procedures in their standard care.[13]

Cytogenetics has the potential to revolutionize Pakistani healthcare by enhancing the precision of diagnoses and the results of treatment for patients with cancer and genetic disorders. Even though there has been a lot of development recently, overcoming current obstacles is essential to reaching this potential. Pakistan can effectively address its increasing burden of genetic diseases by utilizing cytogenetics through education, training, funding, and sensitization of healthcare providers. We must come together as a community including physicians,

researchers, legislators, and patients, to push for a stronger cytogenetics infrastructure in Pakistan as we proceed. By working together, require achieving better health results

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