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GENOMIC EVIDENCES FOR THE OCCURRENCE OF LEARNING DISABILITIES

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ABSTRACT: It is a well-established fact strongly supported by various studies that the occurrence of any sort of disorder since birth is a result of some or the other interference with the genetic setup of the child. Whether it is a direct inheritance from affected people in the family or any condition that enhances the already present yet recessive trait in the child. Time and again since the discovery of these learning disabilities it is the area of interest of the scientists and the researchers as to what sort of alterations in the genes cause such abnormalities. Many studies have been conducted to understand the reason behind the occurrence of such abnormalities in the children. It is still a matter of great curiosity that we are still far away from the conclusion that these learning disabilities are due to an alteration in a certain gene or set of genes and the change in the structure and number of chromosomes present in the cell and the exact reason for any such alteration. Time and again relentless research is being carried out to get to the root cause of the disability so that the children suffering from this and their parents are provided help at the earliest and adequate counselling. This paper is an attempt to get an insight at the role of genes and chromosomes in the occurrence of learning disorders in individuals.

KEY WORDS: learning disability, recessive, abnormalities, alteration, genes, chromosomes

1. INTRODUCTION

The most difficult of all the causes is to know the exact genetic cause of a disease or disorder. Since the study at the genetic level is most complicated and costly, it becomes an area of concern for the researchers and scientists. This genetic level study is even more complex than the molecular level; hence it takes a lot of time to come to a genuine and proper conclusion as to what is happening at the genetic level if a disorder occurs in an individual. The molecular level study for the behavioural problems amongst the LD group is a matter of great research. Recent advances have been made in the area of molecular genetics that have made the study of linkage analysis a very powerful tool for studying the genetics of the behavioural phenotypes of the persons suffering from LD. (Pennington& Smith,1988).

Recent studies in the area of genetics of learning disabilities have concluded that the diagnosis for the genetic diseases is quite different from the traditional diagnosis in a way that the effects of the relevant genes are seen to be largely general rather than being specific. The researches have suggested that the genes that are common to learning disability are general to language impairment, reading disability and mathematical disability rather than being specific for any one. (Plomin,& Kovas,2005), (Haworth & Plomin,2010).

2 to 3% of the population of people with learning disabilities constitute people with poor brain development, the problem apparently more prevalent in the males. Research led by eminent scientists at the Wellcome Trust Sanger Institute, Cambridge, has added 70 other genes on the X-chromosome that have already been found to be directly linked to the abnormal brain development in the boys. In the females the faulty genes on the X-chromosome become counteracted by the healthy copy present there hence such girls become carriers of the disease. (Sample.I.2009). Children who suffer from such learning disability become problem behaviour wise. There are many slow learners who suffer from emotional as well as learning disabilities but can be treated with the process of chemotherapy to some extent which is long lasting up to around six months. (Rossi,1972).

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The presence of learning disability can be result of alteration in either the autosomes or the sex chromosomes of the cell. There can be a part that is missing or an extra number of chromosomes may be present.

2. ABNORMALITIES FOUND IN AUTOSOMES

These abnormalities in the children do not involve the sex chromosome X and Y. These occur when there is any sort of discrepancy in the autosomal set of chromosomes of the cell. These chromosomes are the set of chromosomes from number 1 to 22 which do not take part in the sex determination of the child hence called autosomes.

2.1 Down's Syndrome: One of the most common occurrences of learning disability is with the people suffering from Down's syndrome or the trisomy 21. In this condition an extra chromosome 21 appears in the nucleus of the cell and causes this syndrome. People with this condition are quiet recognisable and the clinical features are quiet profound. In around 8% of the cases of these occur due to the translocation of a part of the chromosome 21 onto another chromosome. This condition occurs in every 650 births. The maternal age is a big factor as mothers above 45 years and also very young ones are noted to give birth to children with these defects. Other trisomy syndromes include the Edward syndrome (trisomy 18) and Patau syndrome (trisomy 13) but their occurrence is relatively low. (Rittey,2003)

2.2 Cry Du Chat Syndrome: This happens when a piece of chromosome 5 remains missing. This is a rare genetic condition occurring in the children. In this disorder the cries of the child becomes very high pitched like that of a cat. The different typical characteristics of this disorder includes microcephaly, low set ears, wide spaced eyes and moderate to severe learning disabilities among the children.

3. ABNORMALITIES FOUND IN SEX CHROMOSOMES

These types of abnormalities occur when the problem involves the sex chromosomes in the cell that is X and Y chromosome. It may happen that any of these remain missing or any one may remain duplicated in the cell. The absence or presence of any of these chromosomes becomes a serious disorder manifested in different types of syndrome which are characterised by any type or types of learning disability.

3.1 X Linked Learning Difficulty: Studies have revealed that this disorder occurs mostly in the boys. This learning disability can be an X-linked disorder. Boys have been seen to suffer more from this disorder than the girls. Though the exact ratio of the gender proportion is still a matter of discussion, the girls seem to be carriers of the disorder linked to X-chromosome. Scientists are of the opinion that there may be more undiscovered genes that contribute to the X-linked learning disabilities. Also the variants that are expected to lower the frequency of their occurrence will be very difficult and costly to discover. This is a great challenge to implement this and bring it into clinical practise (Stratton,2009).

3.2 Fragile X Chromosome-: This cause stands second in most common cause of learning disability. This disorder was recognised at first due to the presence of a fragile site on the X-chromosome when cultured in a medium deficient of folate. Later it was recognised that this syndrome is caused by a large methylated expansion of a CGG repeat in the FMR1 gene. Due to which there is a loss of expression of FMRP which is an RNA binding protein. It was later suggested that FMRP acts as a regulator of an mRNA transport or translation that is important in synaptic maturation and function. These children in early age show mild to moderate learning difficulty. After the age of puberty they show more profound clinical features. (Rittey,2003).

3.3 Klinefelter Syndrome (XXY): This disorder takes place from the non-disjunction of the XY chromosomes during cell division resulting in an individual having an extra X chromosome. Occurrences are few one in around thousand births. Children suffering from this syndrome have typical clinical features and they face a lot of psycho-social problems. These children are seen to have moderate to severe learning disabilities in them. (Ioannides,2003.)

3.4 Turner's Syndrome (XO): This is a syndrome that effects only the females in the human race. This disorder happens when there is a loss of one of the two XX chromosomes in the cells of the foetus. Incidence of such births is one in every 2500 births. Females having this have short stature and non-functional ovaries. In some cases, females having this syndrome have learning difficulties whereas most have normal intelligence level. (Ioannides,2003.)

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4. GENE ABNORMALITIES

Gene transmission is a process that takes place in the humans through cell division. The transmission of the effected gene on chromosome takes place from one generation to the next causing learning disabilities in the children. The different conditions in which learning disabilities occur may result from changes in the structure of the genetic material making up a gene. These changes may vary greatly including the deletion, duplication, addition, inversion and substitution of the parts of the DNA. The occurrence of gene abnormalities in the children are generally categorised by the mode of transmission of the defective gene to the child from the parent. This can take place either by autosomal dominant transmission or autosomal recessive transmission. (Ioannides,2003.)

Reports have suggested that chromosome 1 is also responsible for such disorder. There have been evidences that show that the short arm has some linkage in transmitting the genetic material for LD. Apart from chromosome 1, short arm of chromosome 2 has also been reported in the transmission of dyslexia in a Norwegian family. Another susceptibility locus on the short arm of chromosome 6 has also been reported to transmit learning difficulty. Long arm of this chromosome has also been identified by some researches to be responsible transmitters. (Fiedorowicz. et al.2001).

4.1. AUTOSOMAL DOMINANT TRANSMISSION-

In this case the transfer of the defective gene takes place through the transmission that is reliant upon only one parent who is the carrier of the defective gene and there is a 50% chance of it occurring in the offspring. Here one of the parents carries the defective gene responsible for the condition and the other one is normal. Through the process of the law of genetic transfer the chances of it occurring in the offspring remains 50% while the other 50% chance remains to be normal for the offspring. (Ioannides,2003.)

4.1.1 Prader Willi Syndrome: This happens when there is a deletion of part of the genetic material on the long arm of the chromosome 15 and this usually originates from the father. This happens 1 in 15000 births and affects both male and females in the same way. These children have growth problems like abnormal growth of testes in the males and suffer from the problem of overeating which when remains untreated causes premature death. Such children when taught show a significant degree of learning disability. (Ioannides,2003.)

4.1.2.Tuberous Sclerosis (Epiloia): This is a rare occurring disorder happening once in every 30,000 to 40,000 births. This is has features like growths on the brains and major organs. A butterfly shaped rash (adenoma sebaceum) will usually appear on the face. Epilepsy is common in children with this problem. 60% of the affected children have some or the other learning disability. (Ioannides,2003.)

4.2. AUTOSOMAL RECESSIVE TRANSMISSION-

In this case the transmission process is dependent on both the parents as both are the carriers of the defective gene. Here the chance of the child manifesting the condition remains 25%. 50% chance remains that the offspring will remain the carrier of the condition and the remaining 25% chances are that the offspring will be normal.

4.2.1.Phenylketonuria: This is a condition that severely affects protein metabolism resulting in raised blood levels of phenylalanine. If this problem is not treated and the normal levels not maintained through diet control, this condition becomes toxic and results in brain damage. This occurs once in every 12000 births. If this problem remains unnoticed and untreated there occurs a lack of pigmentation in the eyes, hair and skin of the child. These children when left untreated develop into persons with severe learning disabilities. (Ioannides,2003.)

4.2.2.Hurler Syndrome: This condition is characterised by the abnormal storage of mucopolysaccharides in connective tissue that makes sure that affected individuals are short in stature and have thick coarse facial features and a low nasal bridges. This happens in every 150,000 births hence is a rare condition. Heart defects are the most profound abnormality in such children. Such children have a great deal of difficulty in learning and may suffer from one or more types of learning disability. These children die at around the age of adolescence.

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5. SUBTELOMERIC EFFECT

There are evidences obtained through the use of FISH (fluorescent in situ hybridisation) that in the selected series of children up to 7.4% with moderate to severe learning difficulty and 0.5% with mild learning difficulty have been shown to have sub-telomeric chromosome rearrangements in their cells. This is a costly method but can be requested once a family has a history of learning disability. These defective rearrangements of the chromosomes at the telomere can also be identified in the presence of prenatal growth failure in some cases. (Rittey,2003).

CONCLUSION:

It is an accepted fact that everything we experience happens at the genetic level. But the study and investigation at this level has not yet become common due to its high cost and non- accessibility to general public. Yet it has been established by the researchers that the incidence of learning disability in children is a play of genes and chromosomes. The presence of proper set of chromosomes with adequate genes and genetic material prevents any disorders from happening. Whereas any discrepancy found in the number and structure of the genetic material and the chromosomes plays havoc with the normalcy of the child being born with any such condition. In the coming times it will become imperative to exactly single out the faulty genes on any chromosome and genetic engineering will be required to stop the transmission of any faulty gene or chromosome.

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