

Autism And Related Syndromes

ABSTRACT

The objective of this review is to sum up that autism is a complicated disorder which develop from union of chromosomal and environmental factors by key discoveries from genetic and epidemiological research department. Specific alleles identification has contributed importantly to autism puzzle. As stated by the centres for disease based control and prevention(CDC), ASD seen to be diagnosed more about boys compared to girls. This neurobehavioural disorder is usually could be seen by age of 3 years. Intense difficulties in social interaction and communication with other people, restricted interests, stereotyped responses to situations and other repetitive morphological patterns of behaviour are symptoms or signsthat are related to autism. Complicated, multigenic interactivity and practicable environmental offerings are to be involved by Etiology. For more distant comprehension of the complicated etiology for autism, research fixating on the consequence of gene bygene interactivities or genetic vulnerability to detrimental environmental challenges can be mentioned. Mouse model system use environmental challenges to divergent gene expression and cell pathology as early in development and supplies a efficient way to experimentally manipulate candidate or constituent genes for autism susceptibility.

KEYWORDS

Autism, Asperger's disorder, Autism spectrum, Pervasive development and disorders

INTRODUCTION

Difficulties with social interaction and communication and by restrictive and repetitive behaviours are characterised by neurodevelopmental disorder called Autism. Autism disease, Aspergers disorder, pervasive's developmental disease, childhood disintegrative disease and Retts disorder are included in pervasive developmental disorders which covers under neurodevelopmental disorder. Children on PDD continuum can be seen from suffering disorders who exhibit triad of autism impairments. The first three disorders of autism spectrum like autistic disease, Aspergers disorder, and also PDD-NOS reflects phenotypically divergently and etiological characteristic compared to rettes disease and CDD also those three are presently called as autisms spectrums disorders(1). There be childhood onset with constellation about including atypical behavioural patterns as well as symptoms which span social interactivity also communication(1). In 1943 Leo Kanner was the first one to describe Autism which is infantile. This person made a report with a survey which included 11 children's who were out of the way concerned with any kind of difference in usual non-social surroundings but also inhabited an inability about relatability other than himself since much early in life. A large number of children who were studied never spoke a single word. Those who could really speak were very unusual than normal in different kinds like pronouns reversal, echolalia, having a tough time with socially used languages also idiosyncratics language(1).Autism studied in majorly individuals shows genetics mutations occurring in recent findings. Brain disorders is the ultimate final destinations of these mutations via causing different kinds about diseases or disorders. The same pattern or same design is not followed by all the mutation's is worth to note. Varied kind of mutation is enclosed. Patients suffering from autism will normally not progress in their upcoming future or stages of development if they are not treated during their childhood itself(2).

OBJECTIVE

The aim of this particular article much is about educating more about autism and its spectrum and also the symptoms that come up with it. It also puts light on disorders that come up with autism in an individual suffering from it.

MAIN TEXT

Autism is very complex, behaviourally described, static disease about brain with an surprising 56% increase in pediatrics prevalences between 1991 and 1997 which is that of high of spinabifida or be it cancer even down syndrome which is of huge worry to presently practicing pediatricians(3). Usually when we say autism which comes under the autism spectrums diseases by which we understand the huge group of developmental diseases which are distinguished by deterioration in three behavioural groups which are as follows as the first one is social interactivity followed by the second one which is communication with language also imaginative approach where as the third one includes the spectrum of interest with other activities(3). Epidemiologic survey conducted for its studies shows that prenatal infectious diseases like rubella or cytomegalovirus with environment related factors like teratogens with prenatal insults along with exposure to toxic layout are taken into consideration for few cases(3). These surveyed outcomes of the studies done are unsuccessful to confirm that MMR vaccine which is measles-mumps-rubella vaccine along with immunization are responsible for increase in the rate of increasing autism patients(3).

limitations or boundaries along with considerable phenotypical heterogeneity in the class of diseases(4). Neither it is in expectation that there would be a single whole gene who is responsible for its corresponding expression even if they may be as many as 15+ genes involved nor there Autistic disease is classified under huge group of pervasives

developmental diseases which takes into consideration the retts syndrome with childhood disintegrative as well as aspergers syndrome along with PDD which is otherwise not really specified(4). 0.6% is the range rate for PDD diseases while 0.1-0.2% is the rate for autism which is suggested according to prevalence estimations(4). There is continuous debate ongoing about their clinical is even single biological marker for autism(4). Even with the monozygotic twins the phenotypical expression if this disease varies widely as well as it is varied individually well as concordance about monozygomatic twins is ver much less than 100% due to which its environmental impressions are also important(4). Using various methods which includes genome linked studies also family with case control candidate gene associated studies, multiple susceptible factors are to be explored(4).

Autism spectrums diseases accumulated in families but it is still unsolved about how the individual is at risk or at what exact extent does this occurs due to factors that are genetically relevant or shared even non shared environment related factors(5). Early twin studies estimation is that the proportion of phenotype variance because of genetic relatable factors is about 90% which makes it one of the most probable inheritable above all the developmental disorder, this is found by family studies along with the fact that ASD's aggregated in the families(5). The focus is predominantly on genetically related factors which is a consequence of etiological research. A twin study which was conducted on a large scale indicates the substantial role about the shared environmental impacts, this indicates that recent twin studies support high heritability. The questions about the relative impacts of genetically relatable factors are raised as a result of family studies which also contribute towards the uncertainty about the etiology of the diseases which is included in the spectrum(5). Well there are considerable limitations on the previously done studies. The reliability is limited when we study the rare diseases such as the disorders which comes under the spectrum this occurs due to the fact that twin studies often have only small samples(5). The raised concerns for the potential bias which is introduced by the population selection is due to the fact that none of the previous studies can represent the prospectively populations based random sample. The limitation in reliability may also be due to restricted follow up given time or possible differences in the etiology for different spectrum disorders(5).

The impact on the growth of the disorders which are under the spectrum which provides an open window towards the biology of this category of conditions, has made such a huge progress which is made possible by finding or identifying the genetic variants(6). The variants which are in relation with the autism related disorders have been discovered in hundreds of different types of genes which are really rare and encloses the whole spectrum of mutations be it from the individual pair alterations till the gain or loss of thousands to millions of base pairs(6). The rate of denovo genetic variants which is that the variants who are recognized for the first time in the proband also who are not present in the genome of the parent is increased this is shown by numerous studies in addition to inherited variants in individuals who have one of the disorder which comes under the spectrum(6).

Difficulty in social interactivity with restricted, repetitive design of behaviour or be it interests even if its activities these are the main diagnostic features of children which comes under the autism spectrum(7). The continuum of severity with functional impairment is the reason for the diagnostic criteria to exist. Between six to twelve months of age there may be some signs and symptoms that may emerge where as in many cases a definitive and reliable diagnosis can be made around twenty four months of the age. Most distinguishable characteristics in children who are younger than three years of age are social difficulties as well as delay in response(7). Ability to coordinate individual own attention between another person and an object placed distantly to share interest is recognized as joint attention(7).

Response at twelve months of age and initiation by fourteen months of age towards joint attention can be seen in neurotypical children. The patients should be evaluated who are on the spectrum. If the children who are on the spectrum does not respond even after multiple attempts in order to get their attention by calling out their names then the corresponding patient parents might present with a huge concern of hearing loss(7).

Children who might need a more thorough diagnosis can be more easily identified by screening tools. Better than to rely on clinical judgement, formal screening is more effective(7). Estimating the effective ability of screening for patients on the spectrum in children who are at maximum three years or younger than that based on long term product, there seem to be no randomized clinically related traits(7). There was insufficient evidence about making a recommendation for screen in for children who are not on the spectrum also whose so ever age comes between eighteen to thirteen months of age, this was found out by the United States Preventive Service Task Forces and the American Academy about Families Physicians. Around the age of 9,18,24,30 months the routine development screening has been suggesting to go for well children visits. There has been a recommendation for targeted screening for the patients who are on the spectrum side by side with validated screening tools for early identifying about the disorder which is favourable to perform around the age of eighteen to twenty four months of age, was given by the American Academy of Pediatrics. The most widely useable screening tool is the (M-CHAT) which is the modified checklist for autistic toddlers(7). It has not totally up to the mark positive predictive position also has a high pseudo positive rate, when its used alone(7). The modified checklist for autistic toddlers is a 2 stage parental reported screening tool to estimate the risk of the child who is on the spectrum(7).

Autism is related to fad, unsupportiveness with controversies unvalidated treatment also disprovenness this can be seen while treating the individuals who are on the spectrum(8). Children or even for the adult eclecticism is not a good approach in concern of treating as well as educating them(8). The factors which are identified by none other than the United States National Research Council as the trait of effectivity intermentions in programs related to education with the treatment for individuals who are on the spectrum, the methods derived from scientific studies with its known principle of behaviours incorporate the given above factors(8). For the treatment of the general symptoms like the deviating behaviour of the patients who are on the spectrum for them the applied behaviour analysis is a primary method for its treatment(8). Only on the basic principles of applied behavioural analysis the comprehensive with lasting results for those who are on the spectrum these are the only interventions that is shown(8).

The disorders which are included in the autism related disorders are not rare anymore thus there are many pediatricians who cares or take cares of such individuals(9). The usual first point for contact of the parents of the patients who are on the spectrum is pediatricians who help in early identification of spectrum diseases thus plays a very crucial role(9). Due to the more than often coverage in media about the disorders on the spectrum along with if there is showcase of any of the given symptoms by their child, thus the parents are more aware now about the symptoms that can be seen since a very early age eventually after this they will probably go to their pediatricians along with the issues ongoing(9). Recognition with strategizing of the symptoms for autistic patients also with assessing the accordingly respectively is very crucial for pediatricians(9). The awareness about the local resources which can contribute in process of a definitive diagnosis should be in pediatricians about the spectrum disorders in patients(9). To be familiar with the community or related to education

even the developmental resources along with medical susceptibility clinic, must be done by the pediatrician(9).

Because of the increasing media coverage with a rapid expandable body of knowledge which is published in journals on the professional level, the public and physicians awareness of autism disorder has been remarkably increased(9). The term “autism spectrum diseases” have been introduced by the professionals who are been specialized in the autism sector which from the past two decades is been increased remarkably, the terminology reflects the wide range of clinical traits which nowadays can be identified as autism. As defined in the diagnostic and statistical manuals of mental disorders, 4th edition and in the newest diagnostic and statistical manuals of mental disorders, the text revision of the 4th edition the autism spectrum disease shows around three of the pervasives developmental diseases which is the autism then the aspergers syndrome with the retts syndrome and non- specified PDD-NOS(9). Autism has huge variability in light of presence of the intensity of symptoms even within the diagnostic and statistical manual of mental disorders 4th edition in the text revision groups which indirectly means that there would be additional subtypes, this is in addition to being a disorder in the spectrum series(9). The proper etiology is still not known of the disorders on the spectrum of disease though they are neurodevelopmental situations with a strong genetic underpinnings(9).

The first description of autism was given in a very small group of children who established a very extreme unresponsiveness when compared to non-autistic persons, this was given at the time of 1943 by psychiatrist named Leo Kanner who worked at Johns Hopkins University. An article was published around the corner of 1944 about the children who did demonstrate signs very identical to those of the kanner patient with only the difference of that the cognitive and the verbal skills were higher, this article was by Hans asperger who was an Austrian peadiatrics also unaware of kanner work(9). In the diagnostic and statistical manual of mental diseases there was the first appearance of a diagnostic label of the terminology “infantile autism”. Very much from that time the term has been changed along with the fact that the diagnostic criteria has widened(9-15).

CONCLUSION

The sufferings from the diseases on the PDD continuum can be readily seen in the patients who are on the spectrum also who are going through the autistic impairments. The well higher leve functioning category is less well served where as the diagnostic and statistical manual of mental disorders the 4th edition do identify a low performing autistic group. The PDD-NOS sub group is not in functioning as well as there not remarkable distinguishable factor about the asperger syndrome. Also autism must be start treating since early age to avoid huge reactive setbacks in individual if he is treated afterwards.

COMPETING INTERESTS DISCLAIMER:

Authors have declared that no competing interests exist. The products used for this research are commonly and predominantly use products in our area of research and country. There is absolutely no conflict of interest between the authors and producers of the products because we do not intend to use these products as an avenue for any litigation but for the advancement of knowledge. Also, the research was not funded by the producing company rather it was funded by personal efforts of the authors.

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