

# Autism in Children: Clinical Features, Management and Challenges.

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## ABSTRACT

**Background:** The prevalence of autism world-wide is estimated to be between 1 per 91-110 children. Awareness in the USA and many other developed countries is relatively high and increasing, information on this condition in Nigeria among the public and the medical community is sparse.

**Objective:** This paper highlights the clinical features and management of autism. It also discusses current challenges of autism and its management.

**Method:** Data was sourced from literature in world health organization (WHO) and published articles on autism and internet Medline publications.

**Results:** Autism is characterized by impaired social interaction, problems with verbal and nonverbal communication, unusual repetitive or severely limited activities and interests. The impact of these behaviors can range from mild to disabling. Autism affects all races, ethnic groups and socioeconomic levels, the exact cause is unknown and there is no cure for it. Management is multidisciplinary including educational therapy, psychotherapy, drug treatment amongst others. The combined problems of malaria, malnutrition, pneumonia, HIV/AIDS and other infectious illnesses may be masking emphasis on this neurodevelopmental condition.

**Conclusions:** Autism is a common neurodevelopmental disorder worldwide. Management of the condition is challenging. Creating awareness, improving skills in its diagnosis and optimal management will promote better outcome in affected children.

**Key Words:** Autism, features, poor awareness, management, challenges

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disorder has increased dramatically since the 1980s this was linked to changes in diagnostic practice<sup>1,4</sup>.

Autism affects many parts of the brain; how this occurs is poorly understood. It affects all races, ethnic groups and socioeconomic levels, the exact cause for each individual affected with the disorder is indeed unknown and there is no cure for it<sup>5</sup>. Few children with autism live independently after reaching adulthood<sup>6</sup>. This paper highlights the clinical features (characteristics), proposed causes, management and outcome of autism. It also aims at discussing the current challenges of autism in developing countries like Nigeria.

**History:** Autism was coined from the Latin word *autismus* by the Swiss psychiatrist Eugen Bleuler in 1910 as he was defining symptoms of schizophrenia<sup>7</sup>. He derived it from the Greek word *autos* (meaning self), and used it to mean morbid self-admiration, referring to "autistic withdrawal of the patient to his fantasies, against which any influence from outside becomes an intolerable disturbance"<sup>8</sup>. The word autism, however first took its modern sense in 1938 when Hans Asperger of the Vienna University Hospital adopted Bleuler's terminology "autistic psychopaths" in a lecture in German about child psychology where he described children that played alone and were not interested in their environment<sup>9</sup>. Thereafter, Leo Kanner of the Johns Hopkins Hospital first used autism in its modern sense in English when he introduced the label 'early infantile autism' in a 1943 report of 11 children with striking behavioral similarities<sup>10</sup>. Almost all the characteristics described by Kanner on the subject, notably "autistic aloneness" and "insistence on sameness", are still regarded as typical of the autistic spectrum disorders<sup>11,12</sup>. Kanner's use of autism led to decades of confused terminology like "infantile schizophrenia". Starting in the late 1960s autism was established as a separate syndrome by demonstrating that it is lifelong and also thought to be a heritable psychiatric condition<sup>13,14</sup>.

## Characteristics

Autism is distinguished by a pattern of symptoms rather than one single symptom. The main characteristics are impairments in social interaction and communication, restricted interests and repetitive behavior. Other aspects, such as atypical eating habits are also common but not essential for diagnosis<sup>15</sup>.

## Social development

Children with autism have social impairments. This becomes apparent early in childhood and continues through adulthood. Autistic toddlers have more striking social deviance; for example, they have less eye contact and anticipatory postures and are more likely to communicate by manipulating another person's hand<sup>14,15</sup>. Three- to five-year-old autistic children are less likely to exhibit social

## INTRODUCTION

Autism is a pervasive developmental disorder which results in profound impairment of social interaction, communication, restricted and repetitive behaviors<sup>1,2</sup>. Symptoms usually start early, usually before three years of age. Its cause is unknown but proposed causes include genetic inheritance, teratogens and childhood vaccines. The vaccine hypotheses have been controversial and lack convincing scientific evidence<sup>3</sup>. Most recent reviews estimate a high prevalence of one case per 91 to 110 persons with male-to-female ratio of 4.3:1<sup>1</sup>. Recently, it has been perceived that the number of people affected with the

understanding, approach others spontaneously, imitate and respond to emotions, communicate nonverbally, and take turns with others. However, they could form attachments to their primary caregivers<sup>15</sup>. Making and maintaining friendships often proves difficult for them. There are few reports of aggression and violence in some of them<sup>15</sup>.

### Communication

About a third to half of individuals with autism fail to develop enough natural speech to meet their daily communication needs<sup>16</sup>. Communication problems include delayed onset of babbling, unusual gestures, diminished responsiveness, and desynchronization of vocal patterns with the caregiver. In the second and third years, autistic children have less frequent babbling and may stop talking. These children are less likely to make requests or share experiences, and are more likely to repeat others' words (echolalia)<sup>16</sup>.

### Abnormal behaviour

Autistic individuals display many forms of repetitive or restricted behaviours. The Repetitive Behaviour Scale-Revised (RBS-R)<sup>17</sup> categorizes them as follows.

**Stereotype behaviour:** is apparently purposeless movement, such as hand flapping, head rolling, or body rocking.

**Compulsive behaviour** is an intention of someone appearing to follow rules.

**Sameness** is resistance to change or refusal at being interrupted; for example, insisting that an object remain in a particular place at all times.

**Ritualistic behaviour** involves the performance of daily activities the same way each time, such as an unvarying menu or dressing ritual. This is closely associated with sameness and an independent validation has suggested combining the two factors.

**Restricted behavior** is limitation in focus, interest, or activity, such as preoccupation with a single television program.

**Self-injury** includes movements that injure or can injure the person, such as biting oneself. Dominick and his colleagues reported that self-injury at some point affected about 30% of children with autistic spectrum disorder<sup>18</sup>.

It is important to note that no single abnormal behavior seems to be specific to the autistic child, but they appear to be frequent in them<sup>19</sup>.

### Other symptoms

A small percentage of them show some unusual abilities. This could be memorization of trivial issues or exhibiting extraordinarily rare talents<sup>20</sup>. Also, atypical eating behavior occurs in about three-quarters of children with the disorder. Sleep problems occur in about two-thirds of them<sup>18</sup> such as difficulty in falling asleep, frequent nocturnal awakenings, and early morning awakenings. Parents of children with autism have higher levels of stress<sup>21</sup>. This is because they worry about almost all aspects of the child's development and future prospects. Siblings of children with autism report greater admiration of and less conflict with the affected sibling; they have greater risk of negative well-being and poorer sibling relationships as adults.

### Causes

Autism is thought to be an inherited disorder. However, the mutations that increase the risk of autism have not been identified. Typically, autism cannot be traced to a Mendelian

(single-gene) mutation or to single chromosome abnormalities such as fragile X syndrome or 22q13 deletion syndrome<sup>23</sup>. There may be significant interactions among mutations in several genes, or between the environment and mutated genes<sup>24,25</sup>.

All known teratogens related to the risk of autism appear to act during the first eight weeks from conception<sup>25</sup>. Environmental factors that contribute to or exacerbate autism, and important in future research, include certain foods, infectious disease, heavy metals, solvents, diesel exhaust, phthalates and phenols used in plastic products, pesticides, brominated flame retardants, alcohol, smoking, illicit drugs, and vaccines<sup>26</sup>. Although some persons have implicated certain vaccines, there has been no proven evidence about cause and effect<sup>27</sup>. Parental concern about vaccines had led to a decreasing uptake of childhood immunizations and an increasing likelihood of measles outbreaks. Scientific evidence does not support causal association between the measles-mumps-rubella vaccine and autism; there is no scientific evidence that the vaccine preservative thiomersal causes autism<sup>26,28</sup>.

The risk of autism is associated with several prenatal and perinatal risk factors. Such risk factors included advanced maternal age, advanced paternal age, low birth weight, gestation duration, and hypoxia during childbirth<sup>26</sup>. Autism is associated with several genetic disorders<sup>29</sup>. Some of these genetic disorders include Fragile X syndrome, Cohen syndrome, Tuberous sclerosis, Angelman syndrome, Down syndrome, 22q13 deletion and Smith-Lemli-Opitz syndrome. Mental retardation (ranging from mild to profound) is also a risk factor. Boys are at higher risk for autism than girls. The sex ratio averages 4.3:1. Epilepsy, and some metabolic disorders, such as phenylketonuria, is also associated with autism<sup>30</sup>.

### Screening

Most parents notice their children's unusual behaviors by age 18 to 24 months<sup>20</sup>. Some of the early signs for the child to be evaluated by a specialist without delay include: No babbling by 12 months, no gesturing (pointing, waving goodbye) by 12 months, no single words spoken by 16 months, no two-word spontaneous phrases (not including echolalia) by 24 months, loss of any form of language or social skills, at any age<sup>31</sup>.

Screening tools include the Modified Checklist for Autism in Toddlers (M-CHAT),<sup>32</sup> the Early Screening of Autistic Traits Questionnaire, and the First Year Inventory. Screening tools designed for one culture's norms for behaviors like eye contact may be inappropriate for a different culture<sup>33</sup>. In Nigeria these screening tools are not available, making the process difficult and cumbersome.

### Diagnosis

The diagnostic and statistical manual IV, text revised (DSM-IV TR) is used. Autism is defined in the DSM-IV-TR as exhibiting at least a total of six symptoms, including at least two symptoms of qualitative impairment in social interaction, at least one symptom of qualitative impairment in communication, and at least one symptom of restricted and repetitive behavior<sup>34</sup>. Onset must be prior to age three years, with delays or abnormal functioning in either social interaction, language as used in social communication, or

symbolic or imaginative play. Several diagnostic instruments are available. Two are commonly used in autism research: the Autism Diagnostic Interview-Revised (ADI-R) is a semi structured parent interview, and the Autism Diagnostic Observation Schedule (ADOS) uses observation and interaction with the child. The Childhood Autism Rating Scale (CARS) is used widely in clinical environments to assess severity of autism based on observation of children<sup>35</sup>.

A pediatrician commonly performs a preliminary investigation by taking developmental history and physically examining the child. If warranted, diagnosis and evaluations are conducted with help from autism specialists, observing and assessing cognitive, communication, family, and other factors using standardized tools, and taking into account any associated medical conditions. In Nigeria Paediatric neurologists often make the diagnosis based on clinical manifestations. Differential diagnosis for autism at this stage might also consider mental retardation, hearing impairment, and specific language impairment such as Landau-Kleffner syndrome<sup>34</sup>. Clinical genetic evaluations should be done particularly when other symptoms suggest a genetic cause. Metabolic and neuro-imaging tests are sometimes helpful, but are not routine.

Underdiagnosis and overdiagnosis are problems in marginal cases, and much of the recent increase in the number of reported autistic cases may be due to changes in diagnostic practices. It is particularly hard to diagnose autism among the visually impaired, partly because some of its diagnostic criteria depend on vision, and partly because autistic symptoms overlap with those of common blindness syndromes<sup>36</sup>.

### Management

The main goals of treatment are to lessen associated deficits and family distress, and to improve quality of life and functional independence. Different modalities of treatment have been used over time<sup>37</sup>. No single treatment is best and treatment is typically tailored to the child's needs. Early educational interventions have better benefits<sup>38</sup>. Intensive, sustained special education programs and behavior therapy early in life can help children acquire self-care, social, and job skills<sup>38</sup>. Available approaches include applied behavior analysis (ABA), developmental models, structured teaching, speech and language therapy, social skills therapy, and occupational therapy.

Medications are often used to treat associated problems with the common drug classes being antidepressants, stimulants, and antipsychotics<sup>39</sup>. Other alternative therapies and interventions are available<sup>40</sup>, with few of them supported by scientific studies. Some of these treatments are harmful: for example, elimination diets can create nutritional deficiencies unless care is taken, and botched chelating therapy is associated with death in some cases.

### Prognosis

Autism has no cure<sup>16</sup>. The outcome to a large extent depends on early identification and intervention. Some children recover almost completely while others do not. Most children with autism lack social support, meaningful relationships and future employment opportunities. Although core difficulties remain, symptoms often become less severe in later

childhood. Some adults show modest improvement in communication skills, but a few decline. Acquiring language before age six, having IQ above 50, and having a marketable skill all predict better outcome; independent living is unlikely with severe autism<sup>38</sup>.

### Challenges

Many children with this puzzling condition whose cure and causes are largely unknown are not identified early in Nigeria. An estimated 190,000 children in Nigeria may be living with this condition. Autism is treatable via early individualized, intensive behavioral and educational intervention; these modalities of treatment are not readily available in Nigeria. If intervention begins during the critical developmental ages of 1-4 years, the prognoses are usually better<sup>38</sup>.

There is high level of discrimination against children with "unseen" disabilities. Many schools and services are available for the physically disabled children but very few facilities exist for autism and related behavioral and communication handicaps. The special schools reject autistic children.

Another challenge is that, there are no early diagnostic and intervention centres in all the states in Nigeria. This accounts for under-diagnosis, late identification of affected persons and their management. Children with autism often present with nutritional issues. There are cases of lactose intolerance and allergies which are difficult to manage. Food choices are restricted in Nigeria. Those on special diet, such as gluten-free- casein -free diet, rely solely on imported food alternatives to meet their nutritional needs. These food items are very expensive and out of reach for the average Nigerian family.

Treatment is expensive, and more so in Nigeria. A United State study estimated an average cost of \$3.2 million in 2003 U.S. dollars for medical care, extra education and other care. Publicly supported programs are often inadequate or inappropriate for a given child, and out-of-pocket medical or therapy expenses are associated with likelihood of family financial problems<sup>41</sup>.

Another challenge is family disharmony. Members of the society often think autism is as a result of bad parenting. Thus parents usually live in discouragement and denial. In our society it is believed that there are associated supernatural or spiritual undertone, this is unscientific. There is therefore no basis for unscientific therapies for autism. Ignorance is yet another big challenge in our society. It is discovered that some medical practitioners may know some of the symptoms of autism but are not aware that it can be treated. However despite these challenges, a better outcome can be reached if quality of life for these children is improved through early intervention and advocacy<sup>42</sup>.

### CONCLUSION

There is dearth of information about autism in our environment but the problem exists. The combined problems of malaria, pneumonia, HIV/AIDS and other infectious illnesses often mask this neurodevelopment condition. There is need for more information and creation of awareness amongst the Nigerian populace. Parents need to be aware of the early warning signs. Though it has no known medical cure, it is treatable. Early and appropriate intervention can improve a child's prognosis.

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