Chapter 1
INTRODUCTION

Autism, now termed as Autism Spectrum Disorder (ASD) as per DSM-5 classification, is a Neuro-Developmental Disorder (NDD) seen among children, which is characterized by the presence of marked impairment in social interaction, communication and abnormally restricted activities, interests and with presence of repetitive behaviours. Manifestations of this NDD vary greatly depending on the developmental levels and age of the individual. This disorder is also referred to as early infantile autism, childhood autism or Kanner’s syndrome (American Psychiatric Association, 2000).

Autism is a lifelong condition and many of these individuals require institutional care in later life or may be unable to live independently. Studies of adults with autism reveal that the cumulative mortality rate is higher among autistic patients than the non-autistic peers (Schonauer, Klar, Kehrer & Arolt, 2001). Due to the disability prevailing among autistic individuals, the quality of life will be impaired and the burden of disability will have continued impact on parents and the family as a whole. Apart from the monetary loss and the emotional impact caused by the disability of the autistic individual, the long lasting grief among family members is indeed a matter of great concern.

It was in 1943, that Kanner, a psychiatrist, first described autism in a group of eleven children who demonstrated extreme aloofness and total indifference to other people (Kanner, 1943). The term "infantile
autism" first appeared as a diagnostic label in the third edition of Diagnostic and Statistical Manual of Mental Disorders (DSM) (APA, 1980). The clinical criteria are published in DSM, fourth edition (APA, 1994) and DSM-IV-Text Revision (APA, 2000). The criteria of the DSM-IV and TR are essentially the same. The latest criteria for diagnosis of autism has been included in DSM-5.

By definition, the symptoms of autism must be present before the age of three and the diagnosis is usually possible in the second or third year of life. It is included in the category of Autism Spectrum Disorders (ASD) which includes Autism, Asperger disorder, and Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS). The broader category of Pervasive Developmental Disorders (PDD) include above mentioned three disorders and Rett’s syndrome and Child Disintegrative Disorder as per DSM-IV.

Among the PDDs, Autism is the most commonly seen condition and the severity may vary depending upon the impairment in the three domains – communication, social development and behavioural pattern. In some children the verbal skills are well developed, while in others the skills are poorly developed with very little ability or interest in communicating with others, which will characterize these children into high functioning and low functioning autism respectively. Along with this, one can see varied spectrum of presentation from mild level of disease to severe impairment of functions.

Autism is more commonly seen in males and the gender ratio is 3 to 4 boys: 1 girl (Bryson, & Smith, 1998) which means that the condition is seen about 3 to 4 times in males than females.
At present, figures of epidemiological studies on autism in India are not available. As described by Lorna Wing, the incidence reported in a study in Middlesex in UK, and another in Denmark showed that about 4 to 5 children in every 10,000 will have early childhood autism in typical form, with the incidence of three to four times more often in boys than in girls. There has been a documented increase in the prevalence of autism all over the world (Prior, 2003). However, there is increasing evidence that autism is more prevalent than what was believed earlier. Unfortunately there is a dearth of studies on autism in our state and country.

Autism Spectrum Disorders (ASD) have increased over the past 30 years and recent data in the United States indicate that around 5 children per 1000 are likely to be identified as having ASD (CDC, 2006). This increase may be attributed to increased awareness of ASD and widening of criteria of diagnosis that has led to increased identification of these children or may be due to actual increase of the condition. The prevalence of the disease in European countries are even higher – close to 6 per 1000 (Chakrabarti & Fombonne, 2001).

Two population-based studies conducted by Centre for Disease Control (CDC), Atlanta in selected U.S. locations reported ASD prevalence of 3.4 and 6.7 per 1,000 children, respectively. CDC Atlanta, also conducted two nationally representative surveys, the National Health Interview Survey (NHIS) and the National Survey of Children’s Health (NSCH), in which data was collected from parents as to whether their child ever received a diagnosis for autism. Due to similarities in methodology used by the two surveys, CDC analyzed 2003--2004 data
from NHIS and data from the first-ever NSCH to (i) estimate the population-based prevalence of parental report of diagnosed autism in the US and (ii) assess parental reporting of child social, emotional, and behavioral strengths and difficulties and special-health care needs among children with and without reported autism (CDC, 2006). The prevalence of parent-reported diagnosis of autism was 5.7 per 1,000 children in NHIS and 5.5 per 1,000 children in NSCH. Prevalence estimates in the two studies were similar across age, sex, and racial/ethnic populations.

The U. S. Department of Developmental Services reported a 556% increase (Stokstad, 2001) in the prevalence of autism from 1991 to 1997, a rate that is higher than the prevalence rates reported for other pediatric disorders such as spina bifida, cancer and Down syndrome (Filipek, Accardo, Baranek, et.al., 2000). The reason for this alarming increase may be due to widening for the criteria for diagnosis of autism and increased awareness of the disorder among clinicians and early reporting by parents (Fombonne, 2003).

Recent reports show that there is an increased level of awareness among professionals and policy makers about autism. Similarly, there are several reports in the newspapers and other scientific literature regarding this alarming rise of autism. This may perhaps be due to real increase or may be an apparent increase because of several reasons. Enhanced level of awareness among parents and health functionaries, better documentation and clear definition of the condition, early referral for treatment are some of the attributable reasons for this increase.
Even though autism has its origin from birth itself, the full blown symptoms may be noticeable only during toddler period of life. The exact etiology is not known and hence there are many postulations from genetic related causes to environmental causes. Autism could result from more than one cause, with different manifestations in different individuals. Documented causes of autism include genetic mutations and/or deletions, viral infections, and encephalitis following vaccination. Therefore, autism may be the result of genetic defects and/or inflammation of the brain. The inflammation could be caused by a defective placenta, immature blood-brain barrier, the immune response of the mother to infection during pregnancy, a premature birth, encephalitis in the child after birth, or a toxic environment.

1.1 Need and significance of the study

Even though the exact cause of autism is not described, environmental risk factors are proposed as the most important contributing factor for the occurrence of this multi-factorial condition. Many of these risk factors can be modified so as to reduce the incidence of the disease. Review of literature does not point out the proportion of various factors in the causation of autism in India. Hence in this research study the known-modifiable risk factors of autism were studied and analyzed.

Autism is a matter of great concern to the practicing pediatricians since there is sufficient documentary evidence for increase in the prevalence of autism all over the world. Even though there is no documented study showing the prevalence of autism in India, literature
reveals that the prevalence of the condition in western countries is not less than 1 in 150 children and similar prevalence may also exist in our country. It has been reported that there is an increase in prevalence of this condition in India also. The increase in recent reports may be a real increase or increase due to change in diagnostic criteria or increased referral due to more awareness among parents and professionals. Earlier these children might have been categorized as mentally retarded, hyperactivity and related diseases.

Autism is not very rare in the community and pediatricians are the first group of professionals, who will be diagnosing the condition. Similarly, speech therapists, psychologists, psychiatrists, occupational or physiotherapist and general practitioners will also be required to diagnose the condition. Sometimes, parents seek advice of the above professionals due to some other related problems of their autistic children. They may consult the speech therapist for poor speech and language development or consult the psychologist for any sort of abnormal behaviour in their children. Most commonly they seek consultation of pediatricians or general practitioners for some other medical ailments where they diagnose autism.

Early detection of autism in children is of prime importance as there is scope for early intervention. Many studies have proved that there will be improvement if one can offer early intervention. The clinical diagnosis can be supported by administration of tools for identifying autism and also to categorize them, according to the severity of the problem. Further, the availability of simple tools help professionals for early identification of autism. Even though there are
many tools available for screening and diagnosis of autism, they are time consuming and not validated for Indian population. Hence it is proposed to develop and validate a tool for identification of autism among Indian population.

Early detection of autism is very important since it gives a golden opportunity for early intervention, which will help bring back these children to near normalcy. Clinical assessment and diagnosis is the best method for identification of the condition. There are many diagnostic and screening tools for detection of autism. Many of the tools are for the diagnosis of autism like ADOS, ADI-R, CARS and some are for screening the disease in the community like CHAT and M-CHAT. These tools are developed and validated to be used for children of Western countries. But since there are no validated tools for the Indian population, professionals are using the available ones.

Even though DSM criterion is a simple tool for identifying the condition, it can be used only by professionals. The varied presentation of this condition prompts parents to seek professional help of a speech therapist, who may think that it is an isolated case of speech and language delay and may offer only speech therapy. Sometimes even medical professionals may miss this condition due to lack of awareness of this condition. It is in this context, the investigator thought of developing and validating a simple diagnostic tool for identification of autism in hospital settings among children between 2 and 6 years of age.

Disabilities included under the National Trust Act are in fact developmental disabilities caused due to insult to the brain and damage to the central nervous system. This could be due to several
environmental factors which deprive the brain of oxygen before, during or after birth. These disabilities may take the form of autism, cerebral palsy, mental retardation and multiple disabilities. These are disorders only and cannot be categorized as a disease. They cannot be cured by drugs or surgery but early detection and training improves outcome. This is done using the services of physiotherapist, occupational therapist and speech therapist, community based rehabilitation workers and special educators.

It may be noted that Government of India is giving top priority for the welfare of individuals with disability and Government have passed an Act in Parliament in 1995 viz., the Persons with Disabilities (Equal Opportunities, Protection of Rights and Full Participation) Act, 1995. The purview of the Act extends to the whole of India except the State of Jammu and Kashmir. The Act stipulates that “within the limits of their economic capacity and development, the appropriate Governments and the local authorities, with a view to preventing the occurrence of disabilities, shall undertake surveys, investigations and research concerning the cause of occurrence of disabilities; promote various methods of preventing disabilities; screen all the children at least once in a year for the purpose of identifying "at-risk" cases; provide facilities for training to the staff at the primary health centres; sponsor or cause to be sponsored awareness campaigns and disseminate or cause to be disseminated information for general hygiene, health and sanitation; take measures for pre-natal, perinatal and post-natal care of mother and child; educate the public through the pre-schools, primary health centres, village level workers and anganwadi workers; create awareness
amongst the masses through mass media on the causes of disabilities (PWD Act, 1995)

Under the Act early detection of neuro-developmental disability programs are mandatory and every professional should adopt preventive measures for reducing the disability among individuals. It is in this context that availability of simple screening or diagnostic tools for identification of autism may be popularized and be made available with every professional dealing with children with suspected autism.

The National Trust is an autonomous organization of the Ministry of Social Justice and Empowerment, Government of India, set up under the National Trust for the Welfare of Persons with Autism, Cerebral Palsy, Mental Retardation and Multiple Disabilities Act (Act 44 of 1999) to safeguard the welfare of persons with four disabilities – autism, cerebral palsy, mental retardation and multiple disabilities.

Literature search has not revealed any relevant instruments in English or Malayalam language for screening or diagnosing autism among children, specifically designed and validated for the Kerala population. Hence there is a need for the development of such a tool that can be used for screening or diagnosis of children having autism. It has been observed that in Kerala and across the country, many professionals use Childhood Autism Rating Scale (CARS) for the diagnosis of autism which is a tool developed in western countries.

Usually, parents seek the advice of a pediatrician or other primary care physicians for issues related to abnormality of neuro-development like autism, attention deficit hyperactive disorders, mental retardation, speech and language delay etc in their children. But in some cases,
parents themselves seek services of professionals other than doctors for opinion regarding various symptoms their children exhibit. Many a time those symptoms may be the co-morbid conditions of the underlying autism. For example, a child with autism being presented with speech and language delay may be evaluated by a speech pathologist since the parents seek the advice of the professional. In such cases, a formal evaluation of the neuro-developmental status with special emphasis to rule out autism is warranted, in order not to miss such a condition which will help for further diagnosis and treatment.

Considering the above facts, it was decided to construct a valid tool for detection of autism among children of age between 2 and 6 years with suspected impairment in social interaction, communication or behavior referred to various professionals like primary care physicians, pediatricians and speech therapists in clinical settings. A tool with good psychometric properties, fair cultural adaptation would definitely be a boon to hundreds of professionals working with autistic children.

The neuro-developmental disabilities (NDDs) including autism among children have always been a matter of utmost concern but is yet to be considered as a significant public health problem especially in the low and middle income countries including India. The reason may be that attention and resources are focused on more widely prevalent and visible vaccine-preventable childhood diseases, infections, nutritional deficiencies and neonatal issues. It is inferred that 85% of children with NDDs live in low and middle income countries. Paucity of documented data regarding prevalence of NDDs and their risk factors further aggravates the problem. There is also no comprehensive, valid, reliable
and culturally sensitive screening tool for multiple NDDs that can be used in resource constraint environments. Equally important is to identify standard clinical diagnostic criteria that can be applied in conjunction with a screening tool in low and middle income countries by locally trained professionals. The screening system so developed has to be robust enough to collate the most acceptable and practical clinical criteria of various NDDs and estimate prevalence and associated risk factors of NDDs. It is proposed to conduct this study with the aim of highlighting the problem of NDDs among children and facilitating development of interventions that have the potential to reduce the burden of NDDs in India and other low and middle-income countries. Data on prevalence and spectrum of NDDs in the community and potentially modifiable risk factors will be used for advocacy and enable policy makers in rational allocation of adequate resources for prevention, treatment and rehabilitation of the neuro-developmentally disabled. A strong need for clinically useful diagnostic tools in everyday clinical practice with children having autism justifies the significance of the study.

1.2 About the study

Ironically, even though scientific research has advanced immensely no biological test to determine and diagnose autism has ever been formulated. The mental status of autistic children keeps varying from child to child and it can be observed that many of the children may be of normal intelligence and some may be of subnormal intelligence. Various associated problems like visual impairment, hearing impairment,
hyperactivity, epilepsy, sensory dysfunction, behavioural problems are also present as co-morbid conditions.

Autism being a neuro-developmental disorder with impairment mainly in social and language development, the quality of life of these children will be poor. The mental agony of the parents and near relatives due to the burden of this disability is inexplicable. These children are neither retarded nor normal and a small percentage of children could be identified with mental retardation also. There are a few co-morbid conditions associated with autism like epilepsy, refractory errors, hyperactivity, obsession etc. Even though autism is a clinical diagnosis, professionals usually miss to recognize this disorder at a nascent stage.

This will be disadvantageous for the child since the scope for early intervention for autism is denied. Many studies have proved that early detection and early intervention of autism will improve the general condition of the child. This will help in improving the quality of life of child and reducing the burden of disability. Availability of simple, validated and culturally appropriate tools for early identification of autism will help the clinicians in arriving at an early diagnosis.

Even though autism is a disease of multifactorial etiology, only few published studies are available from India with regard to the risk factors. Identification of etiology and the modifiable risk factors will help in planning the preventive strategies of autism in our country.
In this backdrop, this study was undertaken with following objectives

1.3 Objectives

Primary

- To develop a culturally appropriate tool for the diagnosis of autism among children between 2 and 6 years of age.
- To validate the tool against the reference standard – Childhood Autism Rating Scale (CARS).

Secondary

- To study the modifiable risk factors for autism among children between 2 and 6 years of age.