Autism: Understanding basic concepts

The classic features of infantile autism of extreme social isolation, pathological need for sameness and mutism or non-communicative speech were originally described by Kanner in 1943.[1] The diagnostic terminology used to label children with this complex disorder can be confusing. The terms pervasive developmental disorder (PDD) or autistic spectrum disorder (ASD) are now used to describe five distinct disorders (DSM-IV-TR):[2]

- autistic disorder
- Asperger's disorder
- child disintegrative disorder
- Rett's syndrome
- pervasive developmental disorder, not otherwise classified (NOS) – atypical autism.

Diagnostic criteria and classification have evolved over the years. The DSM-IV 2000[2] criteria for the diagnosis of autistic disorder are based on a core triad of symptoms:

- qualitative impairment of social interaction
- qualitative impairments in communication abilities
- restricted repetitive and stereotyped behaviour pattern, interests and activities.

The child who presents with Asperger's disorder clinically has no significant general delay in adaptive skills, language and cognitive development. The hallmark of Rett's syndrome and child disintegrative disorder is marked neuro-regression and loss of previously acquired language, social and motor skills.

PDD can also be classified according to presumed cause. Syndromic autism has a defined cause, such as tuberous sclerosis and fragile X syndrome. The MePCP2 gene mutations have been identified on the X chromosome in about 80% of girls with Rett's syndrome. This has led to the debate over whether the DSM-IV criteria for autistic disorder apply to Rett's syndrome. Idiopathic autism does not have an identifiable cause, but genetic factors are implicated.[3] In most cases of autism there is no known cause, but the second-hit hypothesis proposes that an environmental factor triggers autism in a genetically predisposed individual. In this issue, Moolman-Smook *et al.* (p. XX) discuss the candidate genes implicated in PDD and also highlight the epigenetic phenomenon.

The immune dysfunction reported in children with ASD does not seem to be of major significance in clinical practice. There is no reported increased rate of hospital admissions of children with PPD suffering from opportunistic infections. This also highlights the fact that at present, there is no biological test to confirm the diagnosis of autism.

The burden of the morbidity of PDD in South Africa is largely unknown. However, increased case identification has mirrored reports of an increased prevalence of the disorder in Europe and America over the past decade.[4] The prevalence of autism ranges from 10 to 20 per 10 000 with a male predominance (sex ratio 3 - 4:1). Comorbid cognitive impairment with an IQ below 70 is present in 75 - 89% of children with PDD, while epilepsy occurs in 30%.[5] The old adage that boys with delayed language development will always catch up means that PDD is often recognised late. It is likely that most children with PPD in Africa are not appropriately diagnosed and are languishing at home because of a lack of awareness and intervention services.

Parents generally insist on knowing the likely outcome of PDD in their children. Health care workers should be prepared to break the unpalatable news on the outcome of PDD. The developmental trajectory of children with PPD is varied. Early language regression between 18 and 24 months tends to herald the onset of PDD. Stereotypic behaviours tend to improve with age, while social isolation and language defects tend to persist into adulthood. Of children with PDD, 60% will be completely dependent for care and only 5 - 15% will acquire social and occupational adaptive skills. Lack of speech at 5 years old and an IQ less than 60 with associated epilepsy indicate a poor prognosis for adult social independence.[6,7]

There is no proven scientific cure for autism. However, anecdotal reports by parents of children who have apparently been cured catch the headlines on the internet. The current state of scientific knowledge on the neurobiology of autism implies that a cure would be a miracle and spontaneous remission unlikely.

Managing children with autism in a poorly resourced environment like South Africa is often frustrating. Chronic neurodisability is overshadowed by the huge burden of infectious diseases which have high mortality. Children with autism should have a comprehensive transdisciplinary evaluation that includes cognitive, speech and language, hearing and educational psychology assessments. This will assist with proper educational placement. A medical examination should be done to identify underlying medical conditions. Clinical assessment of the child with autism will determine the need for special investigations such as EEG and chromosomal analysis.

Moolman-Smook *et al.* rightly point out that behavioural intervention is the main mode of therapy in children with autism. However, interventions such as applied behaviour analysis (ABA) and TEACCH (Treatment and Education of Autistic and related Communication handicapped Children) require specialist manpower and infrastructure. The few special schools in South Africa that cater for children with autism are overstretched and tend to be inaccessible to the majority of the children who need then.

Medications, which include risperidone, are indicated for targeted disruptive behaviours and co-morbid conditions. The prescribing clinician should anticipate the undesirable adverse effects of risperidone, such as weight gain. This is especially important in children with cognitive impairment who are not able to control their appetite. There is no scientific evidence supported by well-designed randomised controlled trials to advocate the use of alternative/complementary therapies such as megavitamin supplementation, hyperbaric oxygen therapy, dietary modification, secretin infusion and heavy metal chelation.[8,9] However, clinicians should be sensitive when discussing the use of alternative therapies since most parents will continue to search for answers and a cure. In South Africa, the clinician should be aware of the role of the traditional healer and be prepared to offer rational supportive counselling.

The child with autism often poses a diagnostic and management challenge. Differential diagnoses to be excluded are developmental language disorders and cognitive impairment. The academic curriculum of medical undergraduates needs to include the teaching of neurobehavioural disorders such as

autism. Community health care workers should also be trained to identify and manage children with neurobehavioural disorders. Political will is needed to transform the educational system so that it adapts to the special needs of children with autism. There is an urgent need to stimulate and expand research on autism in South Africa. This will increase our knowledge of the epidemiology, presentation and outcome of autism in children and also enable the proper planning and implementation of early intervention programmes.

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