PROJECT EVIDENCE

PROJECT EVIDENCE for Prevention of Mental Disorders. The project coordinator is Dr Allan Mawdsley. The version can be amended by consent. If you wish to contribute to the project, please email admin@mhyfvic.org

[3] Indicated Programs are those for young people who will inevitably develop mental disorders unless there is preventive intervention.

[3 a] Biological factors

i Brain injury
ii Developmental disorders including autism spectrum
iii Substance abuse
iv Psychosexual and gender dysphoria

[3 a ii] Developmental Disorders including autism spectrum

See also 2a ii Chronic illnesses

It has always been known that humans have a range of abilities and disabilities. Some commonly occurring patterns of disability have been given specific names and studied to explain their biological cause and pathophysiology as pathways to treatment and prevention.

The scientific study involves:

- defining the necessary and sufficient criteria for inclusion in the diagnostic group,
- identifying the biological, psychological and social factors linked to the subject group,
- validating how the factors lead to the disability,
- testing measures to avoid or treat the causative factors.

Sometimes this research identifies a unitary cause and treatment. For example, phenylketonuria is a metabolic disorder caused by a genetic defect leading to an inability to process a dietary amino acid. The build-up of toxic products in the bloodstream damages brain cells resulting in intellectual disability. Early identification by blood testing of newborns can prevent intellectual disability through dietary management. Future scientific advances may enable repair of the genetic defect.

Most developmental disorders are not yet sufficiently understood to fully identify pathophysiology and treatment. Their definition and etiology is still a work in progress. The syndrome of Autism is sufficiently characteristic and widespread to have become the subject of considerable scientific research, but even so has not yet answered the questions. The history of its research serves as a paradigm for other developmental disorders.

History of autism spectrum disorders. Leo Kanner's first description was of a developmental disorder characterised by impairment of interpersonal relationships, delay and distortion of language development accompanied by obsessive sensorimotor stereotypies. The search for specific pathophysiology and treatment depends, in the first instance, on defining the disorder to obtain a unitary sample. With Kanner's criteria, Lorna Wing's Camberwell study showed a prevalence of 2 to 4 per ten thousand children in the population. Later surveys using broader criteria found higher prevalence. Consideration of autism with its predominantly left-hemispheric language impairment and relatively unimpaired right-hemispheric visuo-spatial processing contrasted with Asperger's syndrome predominantly right-hemispheric visuo-spatial processing impairment and relatively unimpaired left-hemispheric language functioning concluded that the two developmental disorders were variants in the same autism spectrum. This, together with a further widening of the inclusion/exclusion criteria has arrived at an estimated prevalence of about one in a hundred children, a far cry from Lorna Wing's early estimate. It is not that Lorna was wrong, but merely that apples and oranges

require different judgment. With the current broad criteria, it is likely that the spectrum contains a family of different, albeit related, disorders.

Search for pathophysiology. Kanner speculated that this may have been due to impaired mother-child bonding, precipitating a decade of suspicion of "refrigerator mothers", the first of many unsubstantiated speculative hobby-horses. Wider population studies later showed this to have been due to sampling bias because Kanner's subjects were predominantly children of Johns Hopkins University staff whose work commitments limited their parenting time. Even so, it took a long time to dispel the myth because it was sustained by a fragment of half-truth in that severe social deprivation such as that experienced by Romanian orphans did produce an autistic-like state.

Another field of speculation prompted by sampling bias has been about intellectual levels, both for the parents and for their children. Initially speculated that highly intelligent parents were more likely to have autistic children, wider population studies have shown that this is uncorrelated but explained by higher assessment referral rates. Again, the identification of savants, leading to speculation that an autistic child has normal or superior abilities awaiting release from the shackles of an illness is equally unrewarding. Population studies show that children with autism have a very wide range of abilities, but the majority have functional impairments that place them in the disabled persons range. The important issue is to ensure that all children have the educational support to enhance and make best use of the abilities they have.

The functional impairments may also be caused by other factors such as the gross brain pathology of tuberose sclerosis, the effects of Fragile-X chromosome disorder, or the previously mentioned gross social deprivation. In the case of autism there is clearly a heavy genetic loading, particularly on the X-chromosome, but no single genetic cause. The genetic loading is exemplified by higher prevalence in families with autistic members, a high prevalence in twins, a very high prevalence in identical twins, and a fourfold disproportion of males as compared to females. But even with genetic predisposition, speculation continues as to whether there are triggering environmental factors.

Arguments for environmental factors have been supported by observation of seasonal variations in incidence but are bedevilled by the natural changes occurring during development prompting speculation that coinciding events are causally linked. Thus, observation of emerging signs of autism coinciding with vaccination led to causal speculation that has only been refuted by large-scale epidemiological surveys. This is not an objection to speculation but against assumptions of correctness before research validation. Similar caution must be shown towards speculation about the role of diet and the enterobiome as factors in autism.

The absence of identified anatomical defects, microscopic tissue abnormalities or biochemical abnormalities should not deter the search for an underlying pathophysiology of autism (or any other developmental disorder). The library of genetic variations and their physiological effects is growing by the day. When effects are known and measurable the gateway is opened for therapeutic interventions.

Functional observations. The following notes are taken from the Australian Psychological Society website.

Social interaction and communication

Social differences revolve around the give-and-take of normal social interactions, non-verbal social communication, and skills in developing, maintaining, and understanding relationships.

Communication skills vary – some children may have little if any speech, while others may have well developed language skills. However, those that do have language skills often find it difficult to communicate effectively. They may say odd or inappropriate things, make blunt or impolite comments, talk about a specific topic for long periods of time with no awareness that others have lost interest, or say things that are not relevant to the current conversation.

Differences with non-verbal communication include difficulty in making and maintaining eye contact and understanding non-verbal communication of others such as facial expressions and hand gestures such as pointing. Some people with ASD seem quite aloof or detached from others, but this is not the case for all people with ASD, with

some being quite affectionate and fond of company. People with ASD do often tune out when others are talking to them, or appear not to listen, especially when the conversation does not involve their favourite topic of interest.

Behaviour and interests
People with ASD can have a very narrow or unusual set of interests, or they may play in a repetitive way. They may know everything about a certain topic and talk about it constantly, want to watch the same television show over and over, or focus their play on a single toy.

They often show limited imaginative play but may use toys and other objects in unusual ways, such as lining up objects, focusing on the spin on the wheel of a toy car, or watching the light shine off various things. Some children with ASD engage in unusual behaviours, such as hand-flapping or rocking, which is usually a sign of excitement or agitation. Many experience difficulties coping with change but do better when they are prepared ahead of time for changes in routine.

Assessment. Assessment should be by multidisciplinary team, covering psychosocial, paediatric medical, psychological, speech and language, motor skills, social skills and tasks of everyday living.

Treatment. Best results are obtained by a program specifically tailored to the capacities of the child to improve skills and communication and social behaviours. Clinicians, educators, carers and family should share information and be consistent in implementation of the program. Early intervention produces best results.

Implications for other Developmental Disorders

Two separate issues need consideration. One is the search for specific pathophysiology and treatment, and the other is best practice management given the current partial understanding.

Best practice management requires an assessment process that ascertains the pattern of functional impairments. This provides the information for an individualised management plan that enhances and optimises the strengths of the child. Careful reporting of assessment findings may identify an underlying pathophysiological disorder. Further research may identify preventive or treatment factors.

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