Chapter 4

A Paediatrician's Approach To The Assessment Of A Child With Intellectual Disability Or Autism

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Introduction

Consultation regarding a child with intellectual disability or Autism Spectrum Disorder (ASD) is a highly challenging process, yet one that can be systematically worked through by using an approach with three dimensions. Focusing on a purely medical approach is one dimension. The second dimension should be aimed at determining whether both the home and school domains have appropriate structure and stimulation that matches the developmental age of the child. The third dimension relies on attaining an understanding of the family unit's emotional welfare. This includes how rules and emotional regulation are managed including welfare emotions of affection and care, and the extent of emergency emotions of depression, anxiety, and anger. This systematic approach usually needs to be conducted over a few consultations, but if forewarned families usually respect the more thorough approach, rather than the rush to prescribe psychotropic medication.

A comprehensive assessment involves a number of challenges. The first challenge of the initial consultation is to develop a rapport with the parents/carers and to at least secure a connection with the child with the disability. That connection relies on the paediatrician demonstrating that he/she is comfortable with the child who may not demonstrate a 'perfect model' of behaviour in the consultation room. The room needs to be prepared to allow the child the scope of exploration without causing any sense of apprehension or hypervigilance on the paediatrician's part. Fragile equipment needs to be secured away, play resources need to be carefully selected, and cabinets locked - all in the hope of avoiding constricting or negative instructions and pleas from all. Free range play, with the parent guiding the child, whilst the paediatrician respectfully shows confidence in their relationship, goes a long way in securing rapport with the parents.

Generally, the paediatrician should try to engage with the child at an early stage, as parents want to see the paediatrician show comfort with their child and his/her disability. Presenting a down-turned hand for the child to reach out to, is a reliable introductory gesture used in conjunction with a calm manner to make welcoming comments or ask simple questions. This enables the paediatrician to observe the child's level of engagement and communication. Even if this fails, the attempt is the part that the parents respect, and makes it easier to start speaking about the child in detail. It also establishes a connection so that the child is less anxious about subsequent consultations. The aim of the initial appointment should be for the family to leave knowing that the paediatrician has respect and empathy for them and that a comfortable line of communication has been opened with them.

It is important that the paediatrician avoids putting pressure on the child that causes distress. If this occurs, one parent can be asked to take the child outside, while the paediatrician continues the consultation with the other parent. This is usually appreciated, especially if it is apparent that the paediatrician has genuinely attempted to connect and keep the environment as calm as possible for the child. The paediatrician should never show frustration, but rather acceptance of the developmental age of the child.

The Initial Assessment Process

Medical History

The standard paediatric history taking and examination should be conducted. However, often a systematic examination cannot be achieved at the initial consultation, but can be a priority for the next stage of the initial assessment process. It is important to note that

when investigating a child's challenging behaviours, there are several important domains that need to be explored as possible maintaining or exacerbating factors.

Ear, Nose, And Throat System

Otitis media is a possible exacerbant of challenging behaviours. The manifestations can be ear tugging, gouging or banging but it can be less specific with head banging, generalised aggression, or distress. It can be difficult to examine the tympanic membranes properly if a child is unable to cooperate and thus this simple diagnosis can easily be missed unless an obvious perforation has evolved. Baseline audiology is a must, particularly if it has been more than 2 years since an assessment.

Sleep problems are an important area of history taking (Cotton & Richdale, 2006). Compromised sleep, particularly during the REM phase, will lead to day time fatigue and a lower threshold to irritability with daily demands and easily precipitated aggression, especially in the afternoons. Establishing effective sleep patterns is possibly the most important therapeutic target for challenging behaviours. Habitual snoring is permissible but obstructed sleep with apnoeas warrants further investigation.

It is important to clarify sleep hygiene including mapping the pre-sleep routine (i.e., caffeine intake via carbonated beverages, aerobic routines, noise levels, stimulation from TV/DVD/computer/console games), approach to bed time, latency to sleep time, frequency of awakenings, and the approaches to resettling. The secondary gains of nocturnal awakening (e.g., co-sleeping, interactions/engagements with family members) need to be asked about as they are potentially sources of positive reinforcement. Behavioural approaches are usually the main approach required however medication

may be a necessary complement. The opportunity to ascertain how the entire family sleep and their level of day time exhaustion follows on naturally.

The degree of work with breathing and possibility of apnoeas needs to be explored, as well as interruptions of sleep from pruritus (related to eczema) or post nasal drip (related to allergic rhinitis). A sleep diary over a 7 day period may be required as usually sleep studies are not tolerated. Lateral airways x-rays and allergy skin testing (for allergic rhinitis exacerbants) are very helpful for determining whether surgical (tonsillo-adenoidectomy) or medical intervention (nasal inhaled corticosteroids or antihistamines) is required.

Gastrointestinal System

When taking a dietary history, the paediatrician should ask about exclusions and excesses of food types. Unless there is an obvious food allergy or intolerance, particularly in the context of family history, excluding certain food items or elimination diets are usually unnecessary. Where the child excludes food types, the history should establish whether the dietary profile is too restrictive to maintain Iron, B12, and Folate stores. This should be formally tested at some stage to ensure that there isn't an underlining microcytic or macrocytic anaemia at hand. There have been reported cases and personal experiences of correction of B12 deficiency allaying challenging behaviours.

Constipation also needs to be excluded as a contributor to the burden of care and mental health problems of the child (Bosch, et al., 2002). Regular soft to firm bowel motions without the components of strain, pain or PR bleeding should be an established base line and maintained aim. It may be difficult for carers to temporarily correlate this

with challenging behaviours accurately, but when it is presented to them as a plausible explanation then there is usually an agreement that it should be a shared objective. At times a diary of bowel motion consistency and habit is required. The choice of laxative or stool softener, or bulking agent is a matter of individual prescribing choice. The trialling of various options should occur until optimal management of bowel habit is established.

Neurological System

Seizure disorders may be a co-morbid diagnosis in this select group of patients. Epilepsy treatment may already be established but it is still pertinent to ascertain both the benefits and harm profile of prescribed anti-convulsant therapy, which may influence sedation, irritability and impulsivity, and may fuel aggressive outbursts (Ring, Zia, Lindeman & Himlok, 2007). There may be a balance between modifying anticonvulsant regimes and adding psychotropic medication to modify the side effects of the anti convulsant regime (Rutecki & Gidal, 2002). It is important to establish a strong line of communication with the co-involved neurologist in such complex clinical dilemmas.

On the other hand, the nature of the seizure itself (particularly partial complex and frontal seizures) may be a sufficient explanation of aggression (Hirsch, 2005). If seizure related activity is a diagnosis that has not been considered, but is strongly suspected based on history, a home video recording of events is worth requesting. This has been found to be an invaluable source of information while working through the possible differential diagnoses and before resorting to specialist investigations of EEG under anaesthetic or video telemetry.

Physical Examination

In The Consultation Room

The physical examination of the child should concentrate on delineating dysmorphic aspects and focal neurological signs. The art of observation will yield much information and can be done while the child is playing or sitting on the parents' lap. The types of observations that can be made are described in Table 4.1.

[Insert Table 4.1. here]

The extent of a hands-on examination depends on the level of rapport established with the child and how confronting/imposing the examination is in respect of possible sensory averting tendencies. This part of the examination may require the parents to gently physically manoeuvre the child into position, rather than the paediatrician. The paediatrician may even need to make the examination into a play scenario, for example, performing a procedure such as measuring blood pressure on the parents first so that the child feels safe.

Hospital Based Physical Examinations

There are situations where physical examination of the child in a consulting room is impossible or unsafe (in terms of exacerbating the child's aggression), despite repeated attempts. In such situations a planned examination in hospital under sedation may provide the opportunity for a thorough systemic examination. Options for sedation include nitrous oxide gas, clonidine, midazolam, ketamine or chloral hydrate.

It is suggested that there be close liaison with an anaesthetist and that this is done within the domains of a hospital. If sedation is unsuccessful then planning for a general anaesthetic may be required. At such a strategic point, as well as your examination, it may be prudent to co-involve a dental review, venesection (if investigations required), and EEG or MRI (if also indicated).

Additional Investigations

Investigative screens for disability often have a low diagnostic yield and should be tailored to the presentation (Cass, Sekaran & Baird, 2006; Moeschler, 2008). Possible considerations for additional investigations are outlined in Table 4.2.

[Insert Table 4.2 here]

Developmental Assessment

This is probably the most important component of the assessment. The aim is to ensure that the behaviours of the child and expectations of carers and other professionals are aligned with the developmental age of the child. This then provides an opportunity to assess the functionality of supports and strategies at home and school. If the child's developmental age is inaccurately rated (under or over) and the approaches of well-meaning adults are 'mismatched' (either under or over), then there is large therapeutic target to aim for.

The best approach is to ascertain the child's abilities in activities of daily living as a reflection of cognitive abilities. This includes obtaining a baseline on,

- Vision and hearing;

- Fine motor, gross motor, and coordination skills;
- Self care skills;
- Communication (verbal/non verbal skills, use of augmentative and alternative communication systems); and,
- How/with whom the child plays and their scope of interests/likes.

The degree of independence, socialisation, and communication are the salient aspects of the developmental assessment that the paediatrician, carers, and other professionals need to be aligned on. In children with problems of social reciprocity or below the developmental age of 2 years, the paediatrician needs to consider sensory sensitivities and preoccupations in play.

Clinical developmental assessment provides a functional test of skills and may identify specific skill deficits e.g., memory, appreciation of time, emotional understanding and reciprocity, emotional and behavioural regulation, capacity for planning and future anticipation, and insight. Review of prior psychometric assessments is helpful and may extend the paediatrician's understanding of the child's development. In most instances, previous assessments will support the paediatrician's clinical findings, observations, and formulation of the developmental profile. Where there are discrepancies, it is generally safer to rely on the clinical assessment and look for further expertise in understanding the difficulties of psychometric assessment. However, if there is a discrepancy raised with the formulation, then a multidisciplinary assessment is recommended.

Formal developmental assessments for the preschool child could include multidisciplinary team observations, Griffiths Mental Development Scales (Griffiths, 1970), Stanford Binet Intelligence Scales (Roid, 2003), and Differential Ability Scales

(Elliott, 1990). For the older child (prior to school entry), standard intelligence tests include the ,Wechsler Preschool and Primary Scale of Intelligence (WPPSI–III, Wechsler, 2002) for ages 2½-7 years, or The Wechsler Intelligence Scale for Children (WISC–IV, Wechsler, 2003) for ages 6-16 years. A handy office tool for developmental screening is the Brigance Inventory of Early Development (IED-II, Brigance, 2004) from birth to the developmental age of seven years. The developmental assessment also provides the basis on which to make the diagnosis of developmental disorders such as ASD and Attention Deficit Hyperactive Disorder (ADHD).

The paediatrician should ensure that there is a dedicated session to developmental assessment (principally by history) and ensure that the parents truly understand the discrepancy between the child's developmental age and chronological age. However, what is often *not* explained well or understood by parents is that there is a 'ceiling' effect of the developmental trajectory after the age of 16 years. For example, a mildly delayed child's independence and academic skills are likely to remain at the developmental age of a 10-12 years old even though his/her temperament settles down in adulthood, and he/she has sufficient skills to competently fulfil most adult roles.

Psychological/Psychiatric Co-diagnoses

Ascertaining the mental health of the child or adolescent as well as the rest of the family, may require separate consultation time with the parents alone. The child may have co-diagnoses such as,

- ASD, with delays in development of social-emotional capacity and adaptability;
- ADHD, leading to impulsive aggression;

- Depression, leading to a miserable or irritable demeanour and in turn easily flared aggressive lash outs; or
- Obsessive Compulsive Disorder or anxiety disorder, leading to anxiety fuelled agitation and aggression.

Some of these may be innate traits while others may be reactive to the environment at hand.

The paediatrician needs to gauge parental exhaustion and possible concurrent depression, which is frequent, often minimised and under treated in this context. This consultation time also provides an opportunity to delineate the existence of co-morbid depression, anxiety and obsessive compulsive disorders. In addition, it allows the paediatrician to ascertain siblings' adjustment to the potentially stigmatising burden and challenge of living with and accepting a sibling with disability.

Parents' or sibling's psychological issues are important concomitants, that may also underlie the challenging behaviours of the child with a disability. Such psychological issues can compromise the quality of family interactions through emotional withdrawal and passivity, or highly expressed emotion such as emotional over-involvement, criticalness or hostility. This style of emotional communication can erode the autonomy or motivation of the young person with disability and affect supervision of play, predictable structure, and access to other relationships and the community.

School/Community Services Perspectives

Establishing an open line of communication with the child's school is required to confirm the extent that problems are context dependent or pervasive. This also ensures

that strategies are shared between parents and teachers, and may assist in achieving consistent results in containing threatening or difficult behaviours.

The paediatrician has a responsibility to raise concerns on whether the child is receiving the appropriate supports at school to match his/her needs. School visits are invaluable, either by the paediatrician directly, or through the community disability services. In more complex cases, the availability of a community case manager can serve as an interface between the office-based consultant paediatrician and other community services, such as occupational therapy for sensory profiling, speech pathology for augmentative communication strategies, and behaviour support or psychology for behavioural strategies.

Conclusion

Assessment of children or young people with intellectual disability or ASD involves a complex matrix of medical, developmental, psychological and environmental dimensions. A well structured, multi staged approach is essential, and several consultations are required for a comprehensive initial assessment. This relatively methodical and thorough approach is necessary before considering the role of psychotropic medication. While psychotropic medication has a definite role in the optimal management of developmental and psychiatric disorders, they should only be considered if other approaches to intervention at home and school have been duly explored.

Table 4.1

Initial Consultation Observations

At the time of meeting /greeting the child:
▪ General nutrition: appropriateness of height/weight ▪ Dysmorphic features of head (asymmetry of shape), face, hands and digits ▪ Bite marks, scars and calluses of the forearm (self injurious behaviours) ▪ Obvious dental caries during a smile (clue to need for formal dental assessment to exclude significant dental pathology as a source of pain) ▪ Symmetry of facial expression (at rest or even when crying) ▪ The nature of cry (hyponasal in adenoidal hypertrophy)
While the child walks around the room and plays during the course of history taking from the parents:
▪ Stability/symmetry of gait ▪ Ability to transfer from lying to sitting to standing positions (Gower's sign) ▪ Symmetry of limb movements (peripheral weakness or cental tonicity changes)
While sitting up in the parent's lap:
▪ Coordination of conjugate eye movements while encouraging the child to visually following interesting play items (cerebellum signs) ▪ Accuracy of the reach and the absence of resting/intentional tremor or past pointing while offering the child interesting play items (cerebellum signs)
While lying back on the parent's lap with upper garments lifted or removed:
▪ Neurocutaneous stigmata ▪ Rashes (eczema as a source of irritation and possible sleep deprivation) ▪ Excoriation marks as a clue to where pain is manifest (e.g., significant

gastritis/oesophagitis can be associated with scratches localised to the epigastrium and lower sternal edge)

- Presence of palmar/conjunctival pallor (iron or B12 deficiency secondary to restrictive diets)
- Subcutaneous fat stores
- Cardiac examination and anterior respiratory examination
- Abdominal examination via inspection for distension (coeliac or chronic constipation) and gentle palpation faecal loading (constipation), hepatosplenomegaly (storage disorder)
- If the child is comfortable, further inspection to exclude hernias, undescended testes and perianal fissures (chronic constipation)

While the parents carry the child or hold the child while squatting to their level:

- Abnormal spinal curvature
- Neurocutaneous stigmata
- Posterior respiratory examination

Leave the following to the very end or for another consultation:

- Measurement of head circumference, weight, and height
- Examination of
  - ears (effusion and hearing impairment)
  - nose and symmetry of nasal air flow (adenoidal hypertrophy as a cause of obstructed /fragmented sleep patterns)
  - throat (tonsillar hypertrophy as a cause of obstructed/fragmented sleep patterns)

Table 4.2

Clinical Presentations And Investigations To Be Considered

Clinical Presentation	Considerations/Investigations
Child with low average to borderline delay alone	▪ Variation from family norms
Child with mild/moderate or above delay with language delay or dysmorphism	▪ Thyroid Functions Tests, DNA for fragile X, Karyotype and (after consultation with a geneticist) possibly transferrin isoforms and Comparative Genomic Hybridization (CGH) array ▪ Specific genetic testing for clinically apparent syndromes
Boys with above	▪ Creatinine Kinase
Child with severe disability	▪ Above plus urine metabolic screen
Developmental regression (including loss of speech skills)	▪ Urine Metabolic screen, Liver Function Tests, serum ammonia, matched serum lactate/pyruvate, guided plasma substrate/enzyme assays, EEG, MRI/MRS, CSF neurotransmitter studies, ASOT and AntiDNAaseB, as well as fundoscopic assessment by ophthalmologist
Child with macrocephaly, microcephaly, head shape asymmetry, focal neurological signs, and neurocutaneous	MRI and MRS

stigmata	
Child with altered seizure pattern	EEG and MRI/MRS
Child with small head	Review newborn screening results, TORCH, Maternal PKU
Child with nutritional concerns	Ferritin, Folate, B12, FBC and film, Coeliac serology and LFT, stool microscopy and culture