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An Educational Resource

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School-Link Initiative, Department of Psychological Medicine, The Children's Hospital at Westmead
www.schoollink.chw.edu.au

Contents

The aim of this Journal is to improve the mental health of children and adolescents with intellectual and developmental disability through enabling academic debate, research and commentary on the field.

Description and purpose

This journal is a vehicle of expertise about mental health information of children and adolescents with intellectual and developmental disability. As a product of CHW School-Link, this journal is supported by School-Link and a collaborative effort with a multi-agency editorial group from the NDIA and NSW Department of Education. We are extremely proud to present these ideas and invite you as authors to help develop this field and the knowledge base to help support children and adolescents.

On our Website:

www.schoollink.chw.edu.au

The website will be playing a crucial role in the information that CHW School-Link can provide to you.

- *The collection of previous and current editions is located there with the ability to download articles separately.*
- *An invitation for contributions can be found on the website with instructions for authors.*
- *Upcoming training at conferences, workshops and other professional development opportunities will be continuously updated.*

Editorial

School-Link Team
The Children's Hospital at Westmead

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Thank you for reading another edition of our journal. We really enjoy your feedback so this edition has been released with our end of year survey. It's a short survey that gives you the opportunity to let us know your needs when working with children and young people with mental health concerns and intellectual or developmental disabilities.

This edition is jam-packed with information from our team in the department of psychological medicine. Tanya Shenoy give us an overview of the NDIS and positive behaviour support. Ellen McBriarty our Occupational Therapist gives an excellent introduction to Sensory Intervention. Judy Longworth, our Senior Pharmacist discusses stopping medication and Dr David Dossetor gives tribute to Lorna Wing in his article.

Last year, the federal Department of Health released the national roadmap for improving the health of people with intellectual disability. I encourage everyone to read it before the end of the year to feel encouraged that our collective hard work is paying off and getting the attention in the right places. There is still much work to do, but this document lights the road ahead.

You can find the roadmap at this web address: https://www.health.gov.au/sites/default/files/documents/2021/08/national-roadmap-for-improving-the-health-of-people-with-intellectual-disability.pdf

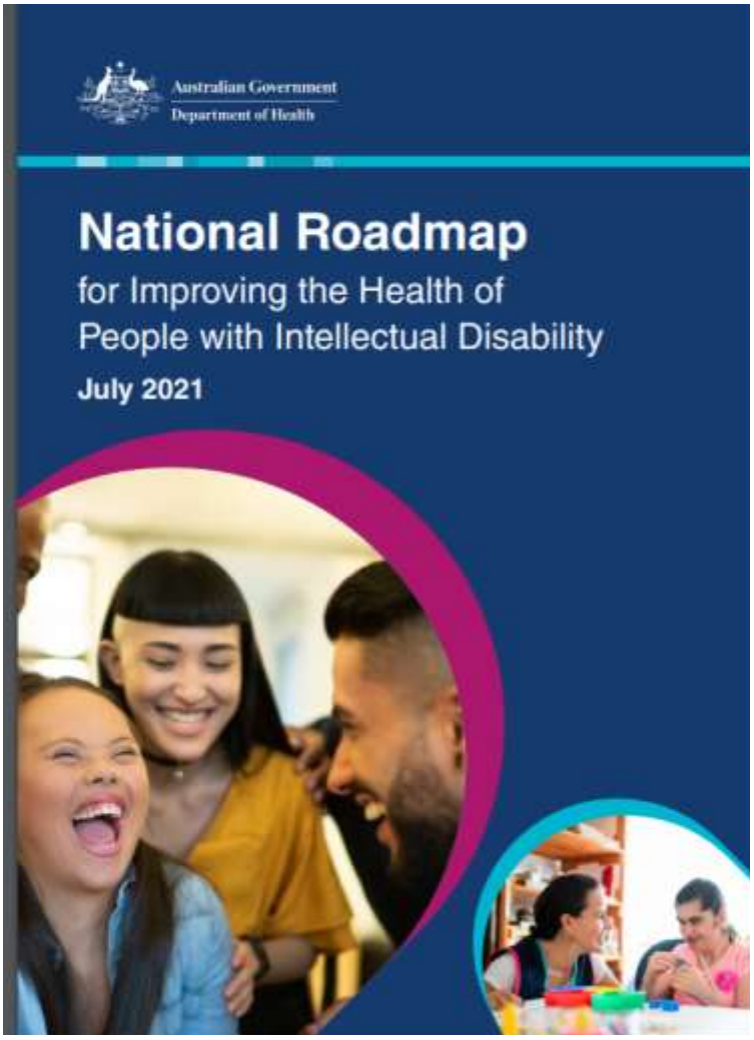
Some important numbers to remember:

Lifeline: Call 131114 or
Lifeline Text: 0477 13 11 14
Kids Helpline: 1800 551 800
Headspace: 1300 737 616
Reachout: Reachout.com.au
Parent line: 1300 1300 52

Enjoy reading this edition of the journal and please send us any feedback by emailing us on the following email address;

SCHN-CHW-SchoolLink@health.nsw.gov.au

Hebah Saleh, Natalie Hunter and Tania Vannitamby
School-Link Coordinators
The Children's Hospital at Westmead
www.schoollink.chw.edu.au



The clinical assessment of development and the implications for understanding social and communication disorders and associated challenges of adjustment. A tribute to Lorna Wing (1928-2014).

Associate Professor David Dossetor

The Children's Hospital at Westmead
Area Director for Mental Health
Child Psychiatrist with a Special interest in Intellectual Disability

Introduction

This article is an introduction to assessing child development as a central skill to developmental psychiatry and understanding the significance of emotions and behaviour in children and in particular those with intellectual disability, autism and other neurodevelopmental problems.

Child development assessment as used for children with intellectual disability and/or autism is often a lapsed skill in senior trainees/clinicians. I find this applies especially to psychiatrists, who either missed that part of paediatric training or have let the detail lapse. With the dominance of adult psychiatry in the provision of mental health services, the significance of a developmental understanding of the mind is customarily overlooked. I find that those who specialise in the mental health of intellectual disability all start with a mental developmental profile as an assumption. When I was training in intellectual disability psychiatry and child psychiatry, I benefitted from re-learning this from Lorna Wing, who along with her clinical psychologist research partner, Judith Gould, wrote the Handicaps Behaviour Skills Schedule in 1980 and whose training I attended in 1985. Although I met Lorna earlier when I trained at the Bethlem Royal and Maudsley Hospitals in London, I came to know her when I became invested in researching families with a teenager with intellectual disability and/or autism. I concur with Judith Gould's description of Lorna as showing "non-competitive excellence".

Quietly, Lorna influenced not just me but the world's understanding of Autism. She developed the concept of an autism spectrum and introduced Asperger's syndrome to the English-speaking world. Lorna's favourite phrase was: "Nature never draws a line without smudging it." That is to say, it is very difficult to draw neat boundaries between those who have and who do not have an autism spectrum disorder. I would expand this to nature often works in dimensions or degrees, but add that development shows sequential progress with time and age.

As a mother of a daughter with autism, she championed the rights of families and proposed that every child should be treated as an individual and that parents and professionals should work closely together. In 1962, she joined a group of like-minded parents to found the National Autistic Society in the UK. In 1991, The National Autism Society's National Autistic Centre for Social and Communication Disorders, in Bromley, Kent, was founded in 1991 and in 2008 was renamed the Lorna Wing Centre for Autism. This was the first place in the UK to provide a complete assessment and advice service for children, adolescents and adults with social and communication disorders. She was passionate about understanding and helping young people with autism. In my mind she did so much through her clarity of thinking, research and teaching.

The Handicaps Behaviour Skills Schedule (HBS) (1982) subsequently evolved into the DISCO (Diagnostic Interview for Social and Communication Disorders) (in which I am also trained) (Wing et al, 2002). However, the DISCO requires 3 days training and takes 3-4 hours to administer and score with an algorithm. In reliability and validity, it is as good as the ADI-R (Autism Diagnostic Interview-Revised), which has the same challenges of luxury of time, and therefore, although either is necessary for gold standard research protocol, I find neither are practicable in clinical practice or indeed in clinical training. The whole of Developmental Psychiatry receives approximately 2 hours lectures in the curriculum for psychiatry registrars, and accordingly the HBS becomes an essential component of self-training and education to be competent to assess young people with intellectual disability and/or autism.

The HBS semi-structured interview is derived from hours of clinical and research interviewing but takes

> “Child development assessment as used for children with IDD is often a lapsed skill ...”

less than an hour to complete. More than that, I have found that completing the interview (re-)trains you in understanding development and with its questions of atypical behaviours, it sensitises you to symptoms which are characteristic of autism. Experience with the HBS gives an awareness of the sequence of skill development that should be inherent to all clinicians that work with these children. Internalising this developmental metric teaches you to adapt your questioning to any presenting individual but also provides an in-depth sensitivity to differences in each domain of skill. An ADI-R is designed to the presence or absence of autism, whereas the HBS (or DISCO) provides the information for a developmental assessment (and therefore a diagnosis of autism) and an individual psychiatric assessment. (Familiarity with the DISCO or ADI-R enables you to cross check a threshold of clinical diagnosis against their algorithms). A few areas of the interview are less central and were designed for collecting data on issues that were teasing Lorna (such as Autism associated neuropsychiatric syndromes, such as catatonia). Lorna was a psychiatrist and a mother of a girl with Autism, as well as the wife of another eminent psychiatrist, Professor of Social Psychiatry John Wing, at the Institute of Psychiatry, at the Maudsley Hospital, who with George Brown described and defined the social factors of schizophrenia and the importance of emotional communication in the relationship with a mental health patient (‘expressed emotion’ was also part of my MD Thesis into families with a teenager with intellectual disability (Dossetor et al, 1994). John was trained in German and translated Hans Asperger’s description for Lorna. Lorna disliked professional arrogance, jargon, fuzzy ideas and unproven treatments and interventions, and was not afraid to say so. Although tough minded, she was, however, always incredibly sensitive to the needs of people with autism and their families. This is the commonsense woman I appreciated.

The HBS was derived from interviews of 150 young people with intellectual disability and autism (Wing, 1980). The semi-structured interviewing enables assessing the reliability of the interviewee by eliciting examples of behaviours that the clinician can evaluate. Of course, no single source of information is adequate for a comprehensive, reliable diagnosis and over-reliance on any one informant has inherent risks. Although the golden rule is that developmental stages generally happen in sequence, neurodevelopmental kids tend to have unequal skills across different domains or have apparent islands of developed skills (savant skills). One example might be an ability to multiply numbers, but not be able to give two objects (i.e. the absence of object number concept). Generally, for adaptive behaviour and ability, the base level skill is more important than the island of advanced skill. Once you have an impression of what level of skill the young person has, one can check this out against the next level of skill above and/or below that identified. However, when you are aware of the general level of skill, one can use questions at a similar developmental level in a different domain.

The HBS Semi-structure interview

Part of the HBS focuses on ability and skills, and part on abnormal or problematic behaviours. The abnormal or problematic behaviours are rated absent, present or severe, based on both frequency and severity (i.e. total impairment). (A sample of HBS interview is downloadable via link).

Each question has an open-ended introductory question such as: ‘How much help does (child’s name) need for dressing?’. Such an opening question can be

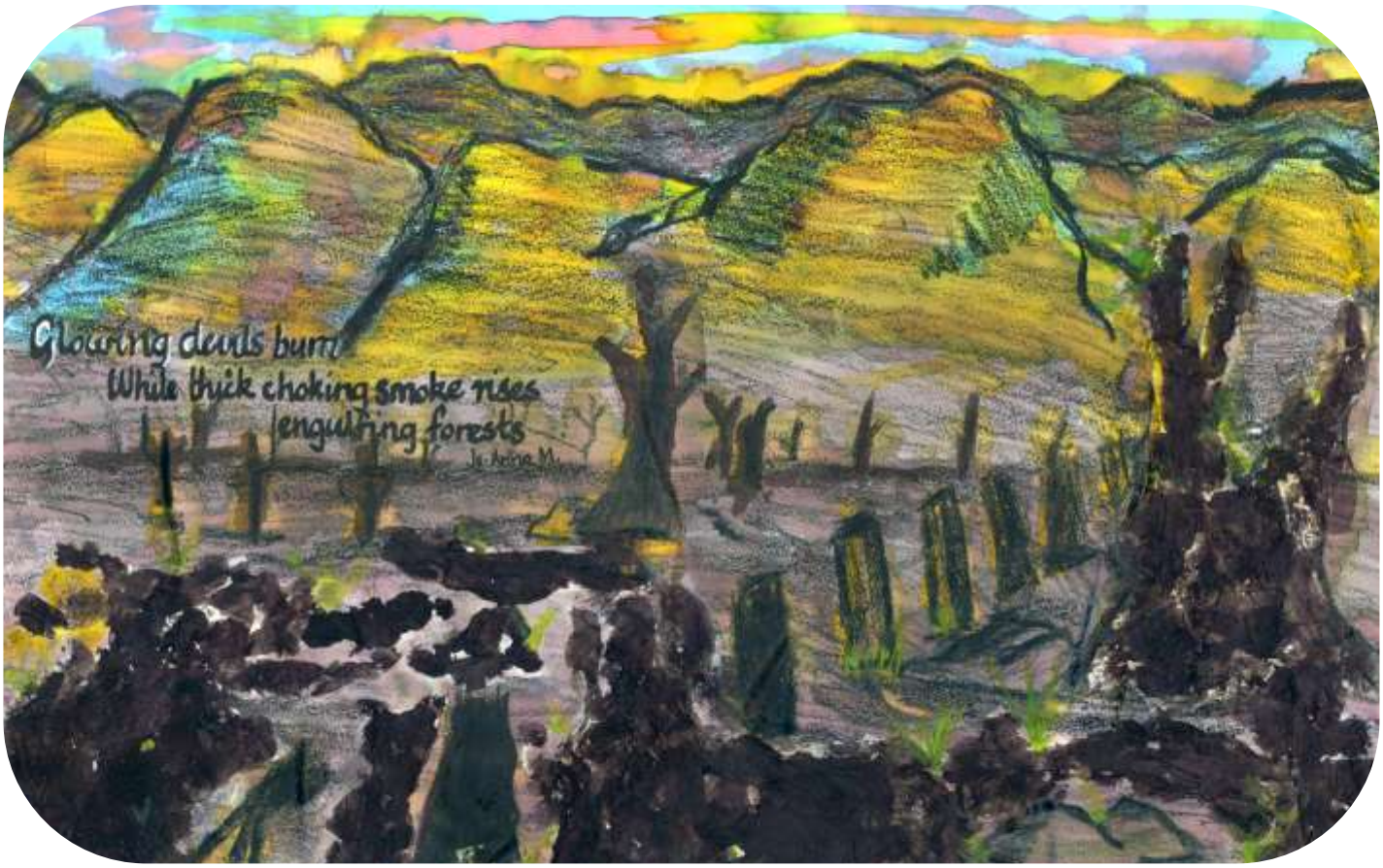
> “The establishment of skills can be influenced by other factors, ”

adapted according to the level of ability of the child. After the open question, clarification can be given with stage-related prompts to elicit appropriate examples. Although self-care skills are the best measure of IQ in those who lack educational skills, the establishment of skills can be influenced by other factors, such as a co-ordination disorder or other problem of motor development such as Hemiplegia. It can also be influenced by parenting style for example where a parent ‘does everything’ for their child. A clinician therefore needs to be aware of such complexities. Many parents declare ‘oh, he understands everything’ and it pays to tie the receptive language question down to ‘what instructions can you give that (child’s name) responds to’ and ask for an example, before defining what developmental age skills are present. For mental health evaluation it is the current functioning that is most important, although the age that a skill developed can be of interest in understanding factors influencing developmental progression, such as the development and control of seizures.

There are two types of abnormal behaviours: First, those that are characteristic of autism and second those that are indicative of abnormal or maladaptive emotions or behaviours. These need to be approached openly, e.g. ‘does (child’s name) tend to have certain phrases or words, that he repeats, which he may have heard others say in the past?’ (delayed echolalia). Such abnormal use of words is rated on the frequency and degree of repetitiveness or duration (e.g. how much of the day is taken up with repetitive behaviour?). Another example is the abnormal response to visual stimuli e.g. unusual interest in shiny objects or the things that spin (an example of abnormal response to visual stimuli).

The second type of abnormal behaviours are things like wandering, destructiveness, tantrums, noisiness, aggression, pestering, rebelliousness, lying or stealing. These can be subdivided into disruptive behaviours with and without social awareness. This distinction on whether a disruptive act such as violence is with insight or intent or a wild lashing-out-of-emotion without awareness of the impact is important in terms of community and professional response and responsibility. The implication is that adults hold a special responsibility for supporting young people with autism who lack insight into their maladaptive behaviours.

There are some areas of enquiry often not considered in similar interviews, such as ability to recognise and communicate facial emotional communication, or



RULES OF DELAYED DEVELOPMENT
1. Assess developmental profile: motor, sensory profile, self-help, receptive & expressive communication, social development, educational and other community skills
2. Behaviour should first be considered from a developmental context
3. If development is delayed, then it is likely to be unevenly delayed.
4. If one domain is delayed, then there is an increased expectation of another domain being delayed. Examples include; If specific language is delayed there is greater risk of Intellectual Disability or problems of social reciprocity If you have coordination disorder then you are more likely to have enuresis If you have delayed development you are more likely to have ADHD 50% of young people with a coordination disorder also have ADHD Autism is more likely in intellectual disability (now confirmed by genetic linkage studies)
Implications: 1. Developmental processes (and impairments) are genetically linked to each other 2. Evaluation of the extent to which an emotion or behaviour is disordered is in relation to the developmental norms and the extent to which they cause additional handicap beyond that expected developmentally.

Diagram 1 Rules of Delayed Development

awareness of time, or how they respond to their image in a mirror or photograph. Or what do they watch on a screen and what is it that interests them, such as movement or music or simple characters or a story. Others include practical skills and avoidance of danger, according to skills of competence. Such questions guide you to appreciating the qualities and individuality of even the most disabled children. Parents so appreciate the skill of 'tuning' into small but critical differences of ability in the severe and profoundly disabled.

In Appendix I below, I provide the main domains of development that the HBS covers to orientate you. Appendix II itemises and illustrates some examples of the mental age-related behaviour and skill sequences. The HBS can be scored to provide a Vineland Adaptive Behaviour Skills score. Appendix III itemises the items of abnormal behaviour (characteristic of autism) and the maladaptive emotions and behaviours which contribute to comorbid mental/psychiatric disorder. Ultimately, the best learning experience comes from trialling the use of the interview and experience and familiarity is what teaches the details. In those who are familiar with the interview, there is a literature describing reliability.

The item of 'Social Awareness and Interaction' is different, as it is not an answer to any item but a clinician's judgement based on both the interview and clinical observations. Social awareness and Interaction is therefore the most challenging and most interesting domain. Wing and Gould (1979) divided social interaction into categories based on their own subdivision of types of social-relating in children with Autism and Aspergers Syndrome:

Aloof: which includes categories 0) 'does not interact, aloof and indifferent' 1) interacts to obtain needs and otherwise indifferent and 2) responds to (or may initiate) only physical contact only including rough and tumble, chasing, cuddles.

Passive: 3) generally, does not initiate but responds to social contact; joins in passively e.g. as a baby in a game of mothers and fathers or for adults. Tries to copy but with little understanding. Shows some pleasure in passive role.
Active but odd: 4) makes social engagement actively but it is inappropriate, naive, peculiar, bizarre or one-sided; behaviour is not modified according to needs, interests or the approaches of the person approached.

Shy: 5) socially appropriate for mental age with well-known people. This may be found in selective mutism, where the subject still communicates with peers, or the withdrawal associated with other psychiatric disorder.

Lastly 6) **Social interaction appropriate for mental age** in both children and adults. (i.e. developmentally appropriate).

Age	Stage	Features
0-1 year	Parent oriented	Development of primary attachment and wariness of strangers. Develop preverbal babble, enjoy rough and tumble. Affective reciprocity.
1-2 years	Adult oriented	Develop capacity for short lived separations; widens range of adult attachments, develop sense of play and humour with adults, such as 'peekabo'. Develop capacity for joint attention. Respond to gross non-verbal emotional communication.
2-2.5 years	Toddler Independence	Copy adults, develop pretend and creative play, become aware of peer play in parallel. Sensitive to subtle non-verbal communication and shame.
2.5-4 years	Peer skill development	Move progressively towards skills of reciprocity with single age-related peer; develop skills of sharing and turn-taking. Initially can turn take if in charge or organised by another. Becoming less ego-centric; popularity comes from organising positive initiatives. Develop first order of theory of mind.
4-8 years	Peer group association	Understand reciprocity to maintain friendship and the practical needs a friend fulfils, eg a friend helps you feel happy. Learn to cope with group relations and social organisations by rules. Second order theory of mind.
9-13 years	Pre-adolescent	Learn to challenge and create group rules. Clear gender split, friendships based on similarity, emotional support, and how they might be viewed by others. Capacity for guilt, sense of object constancy.
13 and older	Adolescence	Based on trust and self-disclosure and mutual or admired aspects of personality. Abstract cognitive capacity.

Diagram 2 Stages of Social Development. Tanguay P, et al 1998 and Dossettor, D, 2004.

Clinical usage and scoring

The HBS provides 2 illustrative scoring systems:
a) developmental ability scoring highlights ability across different domains, what a colleague described as the 'graphic equaliser' of abilities, in the domains of: motor, self-care, language, non-verbal communication, schoolwork, imagination/social and practical skills (attached by link). (Autism is illustrated by the delay in language and social skills behind other skills).
b) abnormal behaviours scores are visually quantified in dimensions of speech, sounds, vision, proximal stimulation, movements, routines, behaviour socially unaware, behaviour socially aware, sleep, and gait, while keeping features of social interaction qualitative.

In clinical practice, I summarise the developmental age of the different domains of motor development, self-care skills, receptive and expressive language and communication skills, educational skills, community independence skills, and emotional/social skills with the family/carers/peers before considering how abnormal the behaviours of concern are in a developmental context. The emotional/social skills I convert with a descriptive scale of developmental ages of social development (Diagram 1.) with an awareness of the rules of development (Diagram 2.) These summaries of assessment also sensitise one to notions of development of the mind (Diagram 3.) (Dossetor, 2019). Meanwhile, one is always also looking for a goodness of fit with other neuropsychiatric/neuropsychological features, such as ADHD, or specific memory problems. Next is the assignment of psychiatric diagnoses, based on the overall symptom pattern, and usually there are co-morbid diagnoses including level of intellectual disability and presence of absence of autism. Further, there may be differential diagnoses, and in particular it can be difficult in autism to determine how much ADHD is predominant over Anxiety or vice-versa (Dossetor, 2019)

Discussion

With their characterisation of social impairment, Wing and Gould were the first to describe a sequence and severity of skills of social interaction that links a progression of severity of autism in this scale to mental age appropriate emotional and social awareness. Others, such as Constantino (Contantino and Todd, 2000) have subsequently confirmed that autism is a dimension of severity and that all severities are all equally genetic. From these observations I have proposed that autism is not just category of loss of reciprocity but is related to a delay in the emotional understanding and consequent social skills (Dossetor, 2004). This sequence of reciprocity skills was described by Tanguay

and colleagues (1991) in his factor analysis of the ADI-R: in year one of life affective reciprocity, joint attention in year 2 and theory of mind in the third year of life. As Tanguay described all these skills are affected in those most affected by severe autism. Problems of theory of mind may be the main deficiency in those less affected.

The work by Dosen and colleagues (Vandevalde et al, 2014) illustrates that emotional development in those with neurodevelopmental disorder is a stronger predictor of emotional/behavioural well-being than chronological age and intellectual level. These observations of the developmental sequence of emotional, and social development, also complement other models of the psychological development viz: Erikson (psychological), Bowlby (attachment), Piaget (cognitive), Bandura (social learning), Kohlberg (moral development), Gardiner (multiple intelligences) and most recently Perry's (neurodevelopmental sequence of the impact of trauma), which are also linked to a model of increasing evolution of the function of the brain and its anatomical substrate, or midbrain/basal nuclear, limbic system, hemispheric specialisation of skills, and

Development of the Mind and Mental Competencies

Mental competencies may be subjective experience, but developmental concepts are critical to understanding children help identify reasons for not coping and showing maladaptive behaviour

- Identification of self and non-self
- Motor regulation and coordination, sensory modulation
- Selective attention and attention switching
- Communication skills and theory of mind
- Mood regulation and empathy
- Self-concept and self-esteem
- Reciprocal social interaction and relationship building
- Reality testing, perspective taking and other executive function skills

Best evidenced by the capacity of a young person to make new good quality peer attachments.

Most important skills:

- Development of attention and concentration, a pre-requisite for learning
- Development of theory of mind: the capacity to appreciate that others have separate thoughts and feelings to your own.

Diagram 3 Development of the Mind and Mental Competencies

"Emotional development in those with IDD is a stronger predictor of emotional/ behavioural well-being than chronological age and IQ"

the development of frontal lobe executive skills (Perry, 2014). Indeed, the other important validated neurodevelopmental disorder ADHD has also shown to be a dimensional quality influenced by environment (Levy et al, 1997). Attentional skills have also been shown to have a developmental sequence (Dossetor, 2004).

Autism was once considered the mental disorder that due to reliability in diagnosis and prognosis was most likely to have a single cause. Now that we know that genetics contribute 40-80% of heritability, with large epigenetic and environmental factors as well (Rylaarsdam & Guemez-Gamboa, 2019). There are approximately 100-1000 different genes involved. Accordingly, we must accept causal processes are multifactorial and differ from case to case. What are the implications for how Social and Communication Disorders should be classified?

In 1982, Michael Rutter led the child psychiatry training group that I was part of and exclaimed that 'the single factor in childhood most predictive of poor mental health in adulthood is poor peer relationships'. This a measure of a primary school aged child's capacity to develop new reciprocal attachments with aged-related peers, which is impaired by both internalising and/or externalising psychological symptoms. This striking observation also identifies childhood social development as key to both child and adult mental ill health or health.

Rutter and Yule (1975) in their landmark paper on reading retardation showed how epidemiology can help classification. Rutter and Yule were able to separate the 2 categories of: 'general reading retardation', where the delay related to multifactorial environmental factors such as IQ, social environment and learning opportunity, and 'specific reading retardation' clusters as a separate condition with correlates indicative of neuropsychological problems.

Similarly, some cases of autism may be a specific disorder of emotional development, due to biological contributors. Other cases can have additional epigenetic and environmental contributors characteristic of 'general delay of emotion and social skills'. For example, general delay in emotion and social skills are associated where the immaturity of social skills are impacted on by other disorders of emotional and behaviour disturbance, i.e. psychiatric disorder.

Russell Barclay's rule of ADHD provides one example: that a child with ADHD is likely on average to be 2 years behind in their social development, based on the observation that ADHD and other externalising symptoms limits their emotional learning opportunity and experience (Wehmeirer et al, 2010). Other examples are the raised rates of autism in severe emotional deprivation as illustrated in Romanian Child refugees, and the lack of theory of mind in deaf children of hearing parents, where there is a lack of exposure to modelling and teaching emotional communication (Peterson &

Slaughter, 2017).

In 1988, I had the opportunity to discuss, with Lorna Wing, the findings of my MD thesis derived from 4-hour interviews of 92 families with an adolescent with severe intellectual disability (Dossetor, 1991). The factor analysis of this study found behaviour disturbance was a single factor. I was surprised that the level of disturbance was not related to chronological age, as it is generally felt that adolescence compounds behaviour disturbance. Instead, behaviour disturbance was related to developmental age, as illustrated in diagram **III.** Lorna responded that the reason is self-evident, since it is in keeping with how a neurotypical child's mind develops. S/he becomes more exploratory, active, risk taking and intrusive in developmental toddlerhood, until the age of 2.5 years when they discover theory of mind, that others have thoughts and feelings separate to them, and they develop an internal world and imagination, into which their mental energy is absorbed, with the corollary of a reduction of the previous externalising toddler behaviours.

The difficulties with theory of mind and social reciprocity is correlated to greater rigidity of thinking and preoccupation with primary processing, as evidenced by stereotypic behaviour and sensory processing issues, possibly as Lorna Wing speculated because this thinking is not socialised.

It was evident that Lorna believed in a model of delayed development of the mind in intellectual disability and autism, rather than a deficit model. In this observation she anticipated the theory and study of the evolution/development of the mind in childhood (Konner, 2012). I find such a conceptual framework of autism of a specific delay in emotional and social learning makes sense to parents, who are able to characterise the age level of social skills in their child, and accordingly describe whether the delay in social skills is in keeping with general IQ or developmental age, or affected secondarily to other psychiatric disorder, or whether the delay in emotional skills inherently delayed behind their other skills. Indeed, those with high functioning autism understand this, in a strengths and weaknesses framework, whereby they may be good at specific cognitive skills in maths, or memory, but have greater difficulty in making friends, like someone much younger.

Further, the Westmead Feelings Program (Ratcliffe et al, 2015; https://www.acer.org/au/westmead-feelings-program) is based on this conceptual framework of understanding autism as a delay in the development and evolution of skills of emotional recognition, perspective taking and problem solving which can be taught as one would for any specific learning difficulty. The research on the Westmead Feelings Program demonstrates that mental health is correlated to emotional social skills, and I hope will contribute to demonstrating sequential emotional skill development in autism.

> "Lorna believed in a model of delayed development of the mind"

DSM V (and ICD11) has focused on a category of ASD, dispensing with the diagnostic categories of Aspergers, Disintegrative Disorder and Pervasive Developmental Disorder but including late onset or atypical autism, and separating ASD from other Social (pragmatic)

> “A developmental framework also suggests that life is a journey”

communication disorders. This has been experienced as stigmatising, as its apparent aim is to exclude all but the most handicapped from accessing support and intervention funding. There remains a shortage of research on the characterisation on the level of needs in other Social Communication Disorders. It also appears to imply that ASD is in some way separate from other Social Communication Disorders. Further, while it may be a form of progress to assess ‘level of support’, there is a lack on criteria on which to rate this, and indeed ‘level of support’ fails to take account of the multidimensionality of problems that contribute to impairment (such as co-morbid psychiatric disorder and level of intellectual disability). Further, there is no metric on which to consider a severity of ASD. DSM V recommended that the severity rating should not be used as a measure of support need, which is exactly what the

The *National Guideline for the Assessment and Diagnosis of Autism Spectrum Disorders* (2018 https://www.autismcrc.com.au/access/national-guideline) now does, despite no research to demonstrate reliability. This categorisation without heed to specific developmental impairment or severity would seem to suggest that taxonomists are looking for aetiological subtypes as a future direction of differentiating ASD. However, it is my surmise that the polygenetic and epigenetic exploration of autism, intellectual disability, ADHD and indeed Schizophrenia will teach us more about the complex normative processes of the development of a healthy mind. Such was Henry Maudsley’s sentiment in 1880:

‘*Anomalies when rightly studied yield rare instruction; they witness and attract attention to the operation of hidden laws or of known laws under new and unknown conditions; and so set the inquirer on new and fruitful paths of research’.* (Henry Maudsley, 1880).

The DISCO (2002) and the ADI-R represent different conceptual/scientific models of examining autism. Lorna Wing was concerned that understanding would be lost by setting a firm boundary around the condition of autism. Perhaps those working exclusively in high functioning autism seek a holy aetiological grail. Those that work with people with intellectual disability ob-

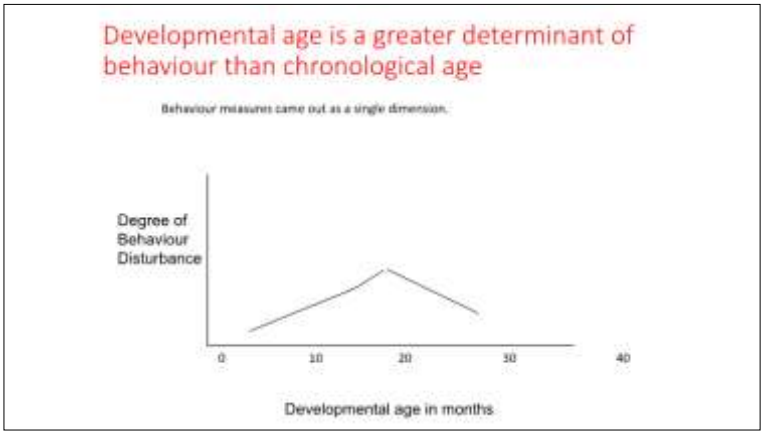


Diagram 4 The relationship between degree of behaviour disturbance and developmental age

serve how overlapping and connected different developmental disorders are, and a developmental framework provide a human based metric that all disciplines, families and the patients themselves can understand, while still looking for salient symptom clusters that characterise psychiatric disorder.

This article represents a key component of training in Developmental Neuropsychiatry. At the time, publishing our multidisciplinary curriculum and textbook (Dossetor et al, 2011) promoting a 'bio-*developmental*-psycho-socio-cultural' framework felt like an approach that unified clinicians across disciplines, as well as families and carers. A developmental framework also suggests that life is a journey, not a destination, and that emotional adjustment, acceptance and belonging remain important for all, no matter where you are on this journey of life and development. I think this is what the human rights and diversity movement articulates.

Understanding trajectory of development helps humanise and make sense of the emotional and behavioural difficulties of young people with neurodevelopmental disorders. There is a compelling literature on theories of development of the mind, and it remains common sense, that development matters for understanding emotions and behaviours. Although this is supported by research findings, the empirical examination of the importance of early emotional and social development is still at an early stage. Expounding the importance of developmental context should not ignore other validated neuropsychiatric models, whether this is the impact of a frontal lobe syndrome or different types of epilepsy, or indeed the genetic or molecular models of behavioural phenotypes.

The HBS is only one of many empirically derived and validated developmental interviews, but it has provided an efficient way of teaching this skill to trainees of neurodevelopmental psychiatry, and I am therefore pleased to make it available to others in the wider neurodevelopmental professional network. The HBS and its developmental framework remains a readily accessible building block in training. While the appendices provide the structure or the interview, nothing beats downloading and using the HBS interview with patients of concern and becoming thereby familiarised. As such Lorna Wing and Judith Gould's contribution continues to familiarise clinicians to neurodevelopmental diversity and autism and provide free access to skills of professional connection with this important population. I thank Lorna Wing and Judith Gould for their legacy which 40 years later provides an efficient assessment, training and education for clinicians concerned for a neurodevelopmentally diverse population.

References

Constantino J, Todd R. 2000. The genetic structure of reciprocal social behaviour. American Journal of Psychiatry. 157, 2043-5.

Dossetor, D.R. (1991). A Study of the dependency needs of adolescents with developmental retardation: an evaluation of short-term care usage, expressed emotion in the primary carer, and the contribution of community services. Doctorate of Medicine, University of Cambridge.

Dossetor, D.R., Nicol, A.R., Stretch, D.D., Rajkowa, S.J. 1994. A study of expressed emotion in the primary carers of adolescents with developmental retardation. Journal of Intellectual Disability Research. 38, 487-499.

Dossetor D. The 10-year celebration of the CHW School-Link and the Journal of Mental Health for Children and Adolescents with Intellectual and Developmental Disabilities. PDF. J of MH for C&A with IDD 2020, 11(1), 4-16.

Dossetor D. 2019. Some personal guidelines for prescribing for the mental health needs of children and adolescents with intellectual disability and/or autism. J of MH for C&A with IDD. 10(1) 4-16. PDF

Dossetor, David. The Evolution of the 'Photon Catcher': Implications for Social Development and Autism. [References]. [Peer Reviewed Journal] *Clinical Child Psychology & Psychiatry. Vol 9(3) Jul 2004, 443-451. Sage Publications, US*.

Gindis B, 2008. Institutional Autism in Children adopted internationally: myth or reality? International Journal of Special Education 23(30): 118-123.

Konner M. 2012. The Evolution of childhood, Relationships, emotion mind. Belnap Press, Massachusetts.

Ratcliffe B, Wong M, Dossetor D, Hayes S. The Association Between Social Skills and Mental Health in School-Aged Children with Autism Spectrum Disorder, with and without Intellectual Disability. 2015. Journal of Autism and Developmental Disorders (JADD): 45(8).

Rutter M, Andersen-Wood, L ,C Beckett , D Bredenkamp, J Castle, C Groothues, J Kreppner, L Keaveney, C Lord, T G O'Connor. Quasi-autistic patterns following severe early global privation. English and Romanian Adoptees (ERA) Study Team. Journal of child Psychiatry and Psychology. 1999 May;40(4):537-49.

Rutter M, Yule W. 1975. The concept of specific reading retardation. Journal of Child Psychology and Psychiatry 16 (3), 181-197.

Rylaarsdam L, Guemez-Gamboa A. 2019. Genetic Causes and Modifiers of Autism Spectrum Disorder. Front. Cell. Neurosci. | https://doi.org/10.3389/fncel.2019.00385

Levy F, Hay D, McStephen M, Wood, C, Waldman I. 1997 Attention-Deficit Hyperactivity Disorder: A Category or a Continuum? Genetic Analysis of a Large-Scale Twin Study. Journal of the American Academy of Child and Adolescent Psychiatry 36(6): 737. DOI:https://doi.org/10.1097/00004583-199706000-00009

Lord C, Rutter M, Le Couteur A, 1994. Autism Diagnostic Interview-Revised: A revised version of a diagnostic interview for caregivers of individuals with possible pervasive developmental disorders. *Journal of Autism and Developmental Disorders* 24, 659–685.

Perry, B.D. 2014 The Neurosequential Model of Therapeutics in Young Children. In (Kristie Brandt, Bruce D. Perry, Stephen Seligman & Edward Tronick, Eds) Infant and Early Childhood Mental Health. pp. 21-47 American Psychiatric Press, NY.

Peterson C, and Slaughter V. 2017 Culture and the sequence of developmental milestones toward theory of mind mastery. Theory of mind development in context. Abingdon, Oxfordshire, United Kingdom: Routledge.25-40

Tanquay P, Robertson J, Derrick A. 1998. A dimensional classification of autism spectrum disorder by social communication domains. Journal of the American Academy of Child and Adolescent Psychiatry 37, 271-7.

Vandevelde S, Morissey F, Dosen A, et al. 2014. The scale for emotional development-revised for persons with intellectual disabilities and mental health problems: development, description and reliability. International Journal of Developmental Disabilities. https://expertise.hogent.be/files/16625471/Vandevelde_Morisse_et_al_IJDD_2014_SED_R.pdf

Wehmeier P, Schacht A, Barclay R. 2010. Social and emotional impairment in children and adolescents with ADHD and the impact on quality of life. Journal of adolescent health. 46: 209-217.

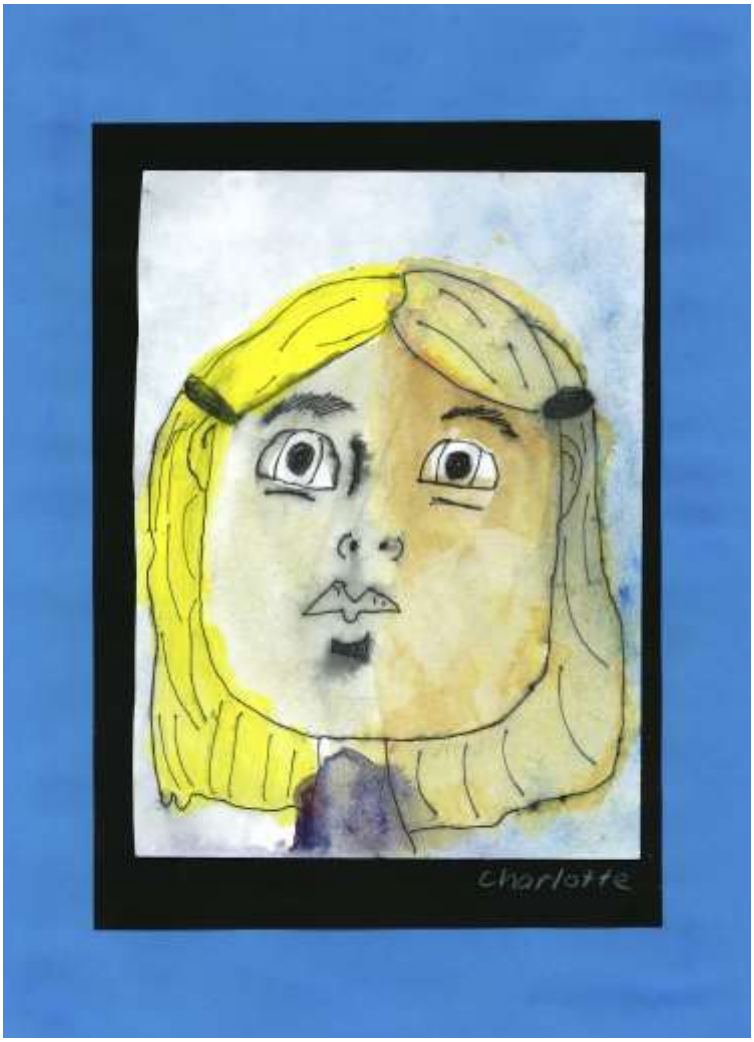
Wing L 1981. A schedule for dividing profits of handicaps in mentally retarded children. In Cooper B (Ed): Assessing the needs of the mentally handicapped. Academic press: London

Wing L. 1982. Schedule of handicaps, behaviours and skills, Unpublished Manual. London: Institute of Psychiatry.

Wing L, Gould J. 1978. Systematic recording of behaviours and skills of retarded and psychotic children. Journal of Autism and Childhood Schizophrenia. 8(1), 79-97.

Wing L, Gould J. 1979. Severe impairments of social interaction and associated abnormalities in children: epidemiology and classification. Journal of Autism and Developmental Disorders 9(1), 11-29.

Wing L, Leekam S, Libby S, Gould J, Larcombe M. 2002. The Diagnostic Interview for Social and Communication Disorders: background, inter-rater reliability and clinical use. Journal of child psychology and psychiatry 43(3); 307-325. https://doi.org/10.1111/1469-7610.00023



Appendix I
The main domains of development of the HBS.

1. **Mobility/Gross Motor Skills,**
   a. Walking on level surfaces
   b. Up and down stairs
2. **Skilled Movements**
   a. Riding a tricycle/bicycle
   b. Manual dexterity
   c. Hand/eye coordination
   d. Problems with coordination/clumsiness
3. **Self-Care**
   a. Feeding
      i. Utensils used
   b. Ability to chew
   c. Drinking
   d. Toilet training
      i. Wee; day/night
      ii. Poo: no supervision needed 3y10m
      iii. Untypical behaviour
   e. Dressing: how much help does A need
   f. Brushing and combing
      i. Buttons,
      ii. Laces
      iii. Untypical behaviours
   g. Hygiene
      i. Washing
      ii. Untypical behaviours
   h. Domestic skills
      i. Tidying, cleaning
      ii. Cookery, woodwork
4. **Independence**
   a. Avoidance of danger
   b. In or out of home
   c. Staying at home alone
5. **Communication (object, visual, sign language, speech, or writing)**
   a. Receptive: general level of understanding
      i. Understanding preposition
      ii. Understanding concepts re future events
      iii. Appreciation of humour
   b. Expressive: ability to use speech
      i. Asking questions
   c. Curiosity
   d. Intelligibility

(Developmental Stages include: meaningful words, 2-3 words with verb, understood by strangers, asking questions; Untypical behaviours: Non-verbal communication, Using a doll/puppet to communicate, Reciprocal communication, echolalia, pronoun reversal, idiosyncratic use of words/phrases, long winded/pedantic, muddling word sequence, problems with making sense, talking to self,)

6. **Non-verbal**
   a. Gestures and miming
   b. Shows what (s)he wants
   c. Joint referencing, sharing interests
   d. Nodding/shaking
   e. Communication to affect others
   f. Describes objects
   g. Emotionally expressive gestures
   h. Facial expression
   i. Body language in social interaction/non-verbal communication
7. **Educational Achievement**
   a. Visuo-spatial with puzzles or building blocks
   b. Use of scissors
   c. 3 d modelling
   d. Drawing
   e. Painting
   f. Colouring in lines
   g. Response to picture books,
   h. Understanding pictures
   i. Response to mirror image
   j. Response to photographs
   k. Money
   l. Days, weeks months
   m. Telling the time by a clock
   n. Understanding of time
   o. Reading
   p. Writing
   q. Numbers
8. **Entertainment**
   a. TV, screentime
   b. Stories read aloud
9. **Imaginative play**
10. **Eye contact**
    a. Social use of eye contact
11. **Social responsiveness**
    a. Show of affection
    b. Response to age-related peers, ability to make friends.
12. **Social Awareness and Interaction**
13. **Practical skills**
    a. Tidying Cleaning
    b. Cookery woodwork
    c. Special skills (above general level)
14. **Initiative and perseverance**
    a. Acquisition of objects ie reaching or managing doors/locks
    b. Spontaneous initiation of activities to do things for self
    c. Nature of chosen activities ie varied and constructive or just repetitive
    d. Attention span for chosen activities
    e. Attention span for tasks given by others
15. **Level of independence**
    a. Understanding of danger
    b. Need for supervision
    c. Staying at home alone
    d. Shopping
    e. Telephone calls

Appendix II Examples of skills and developmental age progression on HBS

Details of all other items is in the interview

Gross Motor Skills

Lift head	4 months
Turns on back	6 months
Sits with support	6 months
Sits without support	7 months
Shuffles or crawls	9months
Walks without support indoors	1yr2months
Walks without support outdoors	1yr5m
Runs more than 45m	2yr 11m
Walks up and down stairs alternate feet	4yr4m
Push a tricycle with feet	2yr 6m
Rides well	4yr

Feeding

Always fed	
Feeds self with fingers	9m
Feeds self with spoon messily	1yr
Feeds self with spoon or fork no help	1yr6m
Feeds self with spoon and fork together	3yrs
Feeds self with knife and fork	5-8yrs
Manage boiled eggs/fish bones	9yr

Dressing

Holds arms/legs out	1yr
Puts shoes on	2yr
Pants down and up and arranges clothes	3yr
Big buttons	3yr6m
Dresses completely but needs clothes arranged in sequence	4yr
Clothes right way round and do all buttons	5yr
Tie shoelaces	5-6yr
Choose clothes appropriate for occasion/weather	12yr5m

Washing

Dries own hands without help	2yr7m
Gets hands acceptably clean and dry without help	3yr 7m
Washes and dries hands and face without help	4-5yr
Baths self without help but with supervision	6yr3m
Can wash and dry hair, cut nails, shave without help	12yr5m

Practical skills eg Tidying Cleaning

Helps a little eg carry cup to kitchen	1yr9mo
Simple immediate tasks eg putting something on shelf	2yr
Fetches or carries to other room or takes message	3yr
Gives help for sequence of actions eg clearing or laying table, dusting,	3yr7mo
Helps regularly without supervision	8yr 6mo
Does some tasks in own initiative for payment	10 yr 11mo
Is responsible for a domestic task eg weeding garden or car washing	14yr 8mo

Tidying, cleaning, cooking, woodwork

Help carry cup to kitchen, help with cooking eg stir cake mix	1yr9m
Help with sequence of actions eg laying/clearing the table	3yr7m
Sew hem, sandpaper wood	3yr6m-4yr
Helps without supervision	8yr6m
Complex task, cook eggs and bacon, simple woodwork	11yr3m

Avoidance of danger/independence in or out of home

Avoids falling from heights	2yr
Avoid danger of traffic	5 yr
Go to local shop without traffic	8-9yr
Left alone at home for 1 hr	10yr
Crossing road safely	10-11yr
Left at home for half day	11-12yr
Left home all day	15yr
Go around nearby town	15yr
Go alone to remote place	18yr

Appendix III Abnormal behaviours on the HBS

The abnormal or problematic behaviours are rated absent, present or severe, based on both frequency/duration and severity (i.e total impairment).

Abnormal imaginative activities

- Stereotyped play or other symbolic activities
- Fantasies (preoccupation with)

Abnormal response to sounds

- Distress caused by sounds
- Fascination with sounds
- Other: ignoring loud or overreacting to almost inaudible sounds

Abnormal response to visual stimuli

- Unusual interest in bright lights and shiny objects
- Interest in watching things spin
- Twisting/flicking his hands or objects near his eyes
- Interest in looking at objects from different angles
- Other response to visual stimuli

Abnormal response to peripheral stimulation

- Mouthing of objects
- Smelling of objects or people
- Touching objects for its feel
- Scratching or tapping surfaces
- Repetitive destructive activities eg paper or bits of wall paper or toys
- Repetitive aimless manipulation of objects (not near eyes)
- Self-Injury
- Self-Stimulation without injury, eg pushing eye, regurgitating food, self induced vomiting, tapping chin, grinding teeth.
- Other repetitive sensory activity

Abnormal response to bodily movements

- Tip-toe walking
- Aimless movement
- Other abnormal bodily movement

Routines and resistance to change

- Dislike of change in normal routine
- Routines invented by the child
- Food fads
- Clinging to objects
- Interest in special objects or part of objects (eg light switches, church steeples, people's teeth
- Special fear: eg dark, big dogs, trains

Behaviour problems involving limited or no social awareness

- Wandering
- Destructiveness
- Noisiness
- Temper Tantrums
- Aggressive behaviour incl spitting
- Hyperactivity
- Behaviour in public places
- Lack of cooperation
- Crying and moaning
- Difficult or objectionable personal habits, eg spit, smear, make vomit, hoard rubbish, inappropriate swearing, inappropriate sexual beh without social awareness.
- Scatters or throws objects around (creates chaos aimlessly)
- Other beh prob with limited or no social awareness

Behaviour problems with social awareness

- Difficulties with other people eg tease, bully refuse to take turns, make trouble
- Rebellious awkward or cheeky behavior
- Pestering for attention
- Lying, cheating, stealing
- Other behaviour probs

Sleeping problems.

- Needs night sedation
- Other disturbance of sleep, eg late to sleep, waking in night, restlessness, noisiness, waking early

Sexual problems: Includes: masturbation in public, inappropriate hetero or homosexual behaviour, sexual interest in much younger children, indecent exposure, other inappropriate sexual behaviour.

Other psychiatric problems

Includes items on: depression, mania/hypomania, anxiety, hypochondriasis, obsessional neurosis eg hand washing, other neurosis, schizophrenia, other psychosis, personality disorder, other psychiatric disorder, abnormalities of mood, organic dementia or confusional state.

Legal problems or status

NDIS & Positive Behaviour Support

Tanya Shenoy
Acting NDIS Manager and NDIS Mental Health Officer
Sydney Children's Hospital Network

What is Positive Behaviour Support (PBS)?

PBS is an evidence-based, person-centred approach which aims to improve a child's quality of life whilst reducing the frequency and severity of challenging behaviours. PBS takes a multifaceted approach to determine the cause and function of challenging behaviours which can be triggered by a variety of factors such as communication difficulties, frustration, avoidance, sensory issues, and more. Through the PBS process, stakeholders such as the child, family, school, and clinicians work collaboratively to develop individualised goals. Further information about the PBS process can be accessed via the Aspect website.

How can a child access PBS?

Funding under a subcategory of Capacity Building known as "Improved Relationships" will be required in an NDIS Plan. The level of funding will depend on the child's individual needs and what is deemed reasonable and necessary (Section 34 of the NDIS Act 2013) to support their circumstances. Behaviour support funding might consist of:

- Therapeutic support in an individual or group setting to help a child build healthier relationships, manage emotions, and improve their overall well-being
- Support groups to learn how to interact with peers and practice social interactions and skills
- Behaviour management training and strategies to support carers in improving their understanding of a child's needs and preferences
- Intensive specialist behaviour intervention to address significantly harmful or persistent behaviours of concern which may involve regulated restrictive practices

How can a Local Area Coordinator or Support Coordinator assist?

If Improved Relationships is not currently funded in a child's NDIS Plan, a parent or legal guardian can request a review. If a Support Coordinator is funded in the Plan, they can assist with the review process, otherwise the family can contact their Local Area Coordinator for assistance.

Ask the family if they have a Support Coordinator and/or check the child's NDIS Plan to see if Level 2 or Level 3 Support Coordination is included (located under the Capacity Building category on the NDIS Plan). Support Coordinators can assist in the following ways:

- Obtain and collate evidence (such as police/hospital reports, support letters, incident reports) to bring to a planning meeting with the NDIS to advocate for behaviour support funding
- Awareness of a child's needs to notice when funding for behaviour support may be required
- Support families to understand NDIS review pathways and navigate restrictive practice processes with a Behaviour Support Practitioner
- Research the local availability of behaviour support providers

How to locate a NDIS Behaviour Support Practitioner

You can use the NDIS Provider Finder webpage or request the assistance of the child's Support Coordinator or Local Area Coordinator.

Some considerations when searching for Behaviour Support Practitioners can include:

1. The practitioner's experience in working with a particular disability, age, and complexity.
2. Will appointments occur at home, in the commu-

nity, at school, or via Telehealth?

3. The relevant goals the child and family would like to work towards. Does the practitioner have the skills and experience to support these goals?
4. Can the practitioner speak a language preferred by the family?
5. The wait time for services to begin.

Please see SCHN's Choosing my Team resource for further guidance.

Working with Families from CALD backgrounds

When working with culturally and linguistically diverse (CALD) families, it is important to understand that cultural expectations, norms and values held by professionals and families can vary and this can influence views on parenting practices, priorities, and expectations. Parents should be acknowledged as the experts on their child and asked about their child's routines, interests, abilities, and needs. Working within an inclusive, strengths-based approach with families can increase engagement with services and a child's connection to their culture, family, and community can be a significant protective factor in nurturing their overall wellbeing.

Families from similar ethnic origins may share some cultural practices, values and beliefs but they are not defined simply by their ethnicity or race; individual characteristics and qualities should be recognised and valued. Avoid assumptions by asking parents about their understanding of behaviour support and offer resources in a preferred language where possible. Some families may be cautious or hesitant about working with new service providers due to negative experiences with service systems or feelings of inadequacy about their ability to provide care for their own child. Avoid an interrogatory style of questioning and allow parents to steer the conversation. Furthermore, communication needs to be clear and accurate, hence if there are language differences, utilise the support of interpreters.

TIS National

NDIS participants and family members who have English as a second language can access fee-free interpreting to implement their NDIS Plans. Translating and Interpreting Services (TIS) National has partnered with the NDIS for this purpose. TIS can supports participants with language interpreting when connecting with registered service providers for funded supports in a

participant's NDIS Plan. A Local Area Coordinator or Support Coordinator can help NDIS participants access interpreting services though TIS National. TIS National requires the NDIS registered service provider who is delivering the funded support to register for a TIS National client code and book an interpreter on behalf of the NDIS participant. The NDIS can also provide a child's NDIS Plan in the family's preferred language. See the language and interpreting services page of the NDIS website for further information or call the NDIS on 1800 800 110.

Resources

NDIS Quality and Safeguards Commission: Regulated Restrictive Practices with Children and Young People with Disability Practice Guide

Understanding Behaviour Support Practice Guide: Young children (0–8 years) with developmental delay and disability

Understanding Behaviour Support Practice Guide: Children and young people (9–18 years) with disability

Autism Spectrum Australia (Aspect)

Raising Children Network

Resources and Readings List

Readings

Eagleson, C., Weise, J., Cvejic, R. C., & Trollor, J. N. (2022). Evaluation of an intellectual disability mental health core competency framework. *The Journal of Mental Health Training, Education and Practice*.

Halvorsen, M. B., Helverschou, S. B., Axelsdottir, B., Brøndbo, P. H., & Martinussen, M. (2022). General Measurement Tools for Assessing Mental Health Problems Among Children and Adolescents with an Intellectual Disability: A Systematic Review. *Journal of Autism and Developmental Disorders*, 1-73

Havercamp, S.M., Barnhill, L.J., Bonardi, A., Chapman, R.A., Cobranchi, C., Fletcher, R.J., Rabidoux, P., Seeley, J.R., Tassé, M.J. and Nisonger Center RRTC on Health and Function, 2022. Straight from the horse's mouth: Increasing self-report in mental health assessment in individuals with intellectual disability. *Journal of Applied Research in Intellectual Disabilities*, *35*(2), pp.471-479.

Lunsky, Y., Jahoda, A., Navas, P., Campanella, S., & Havercamp, S. M. (2022). The mental health and well-being of adults with intellectual disability during the COVID-19 pandemic: A narrative review. *Journal of Policy and Practice in Intellectual Disabilities*, *19*(1), 35-47.

Shield, L., Riney, K., Antony, J., Ouvrier, R. and Ryan, M. (2016). Fifty years of paediatric neurology in Australia. *Journal of Paediatrics and Child Health.* Vol 52, Pp 861-864.

Links

Working with people with mental illness and psychosocial disability
https://www.health.nsw.gov.au/mentalhealth/psychosocial/Pages/default.aspx

Disabled people being 'systematically ignored' on climate crisis, says study https://www.theguardian.com/environment/2022/jun/10/disabled-people-systematically-ignored-climate-crisis-study?CMP=Share_iOSApp_Other

Books

Autism and Me: A book for Children with Autism. By Chris Wever. You can also find his other books for children about Anxiety and Onsessive Compulsive Disorder.
https://www.bookdepository.com/Autism-Me-Chris-Wever/9781922644978

Sensory: Life on the Spectrum by Rebecca Ollerton. An autistic comic anthology exploring a wide range of austistic experiences from diagnosis journeys to finding community. Organised by Schnumn. https://schnumn.com/sensory
Order through Booktopia
https://www.booktopia.com.au/sensory-life-on-the-spectrum-rebecca-ollerton/book/9781524874766.html?gclid=Cj0KCQiAgribBhDkARIsAASA5bumO2YTGv4ZPXaQIt09XSqHO-bAeXnGBRQL5jHP8vE58UD9SEPX50waApCfEALw_wcB

Sensory Intervention in Mental Health and Developmental Disabilities

Ellen McBriarty
Senior Occupational Therapist
Department of Psychological Medicine
The Children's Hospital at Westmead

A wide range of intervention options exist for children experiencing mental ill-health and intellectual and developmental disabilities (IDD). Sensory intervention should be included as much as possible as it is key in developing a secure base in which to increase engagement with daily life and psychotherapies. Research emphasises the importance of focusing on sensory processing in young children in order to understand the characteristics of vulnerable children (Dunn, 2007), such as those with mental health or IDD. This includes building an understanding of how a child's sensory processing may be impacted by:

- early-life events, such as the experience of perinatal, postnatal or early-life trauma, long periods of hospitalisation;
- diagnoses inhibiting sensory learning, including Autism Spectrum Disorder, hypoesthesia, hearing loss or deafness, low vision or blindness or Sensory Processing Disorder;
- environments restricting interaction with sensory stimuli, such as living with or being raised by an individual with restricted sensory processing, or being deprived of the ability to explore, climb, mobilise or mouth items as an infant.

Without this understanding, we cannot begin to plan or implement the ongoing sensory interventions required to assist in regulation.

Sensory development begins in utero with the development of the brain stem, deeply connecting sensory regulation to our survival instincts. Under a mental health lens we see that sensory regulation directly effects our automatic responses (i.e. fight/flight) and forms the base of our ability to process stress, engage in critical thinking and develop attachment in relationships (Lyons et al., 2020). Sensory learning is most intense in the first 12 months of life, as sensory input is experienced, interpreted and understood for the first time.

Children with early life diagnoses of IDD are likely to have a reduced store of sensory memories as their brains may inhibit the interpretation of sensory input (Marco et al., 2011). These children may be less likely to seek new input, and often hold a strong aversion to some sensory input. Similarly, when mental health is experienced by a child or adolescent, neural pathways

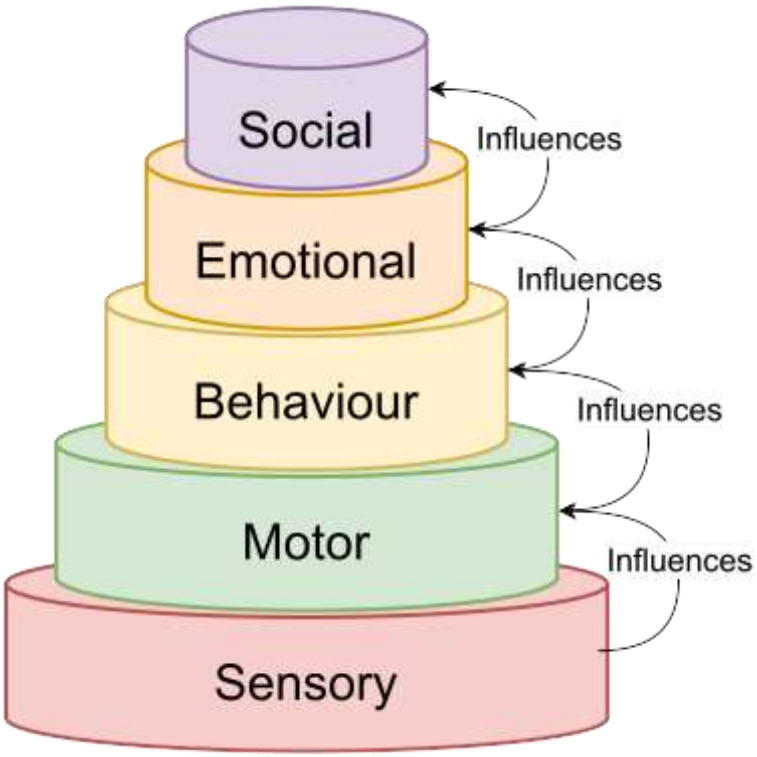


Diagram 1: Relationship of sensory regulation to functional performance.

may be altered in response to stress or sadness, changing their response to specific sensory input (Child Welfare Information Gateway, 2015).

In children with both mental health and IDD, symptoms of reduced, restricted or uncoordinated motor movement, poor social skills or reduced capacity to build relationships and challenging or frequently changing behaviours are common (Camarata et al., 2020). This may be due to our need for secure sensory regulation in order to engage the Limbic and Cortical brain functions of emotional and social development and critical thinking (see diagram 1).

It is well known that children experiencing IDD have altered results on standard sensory processing assessments, indicating a need for targeted sensory intervention before commencing skill building therapies. . Professionals and families working in this space must be equipped with working knowledge of sensory processing in order to accurately interpret children's behaviours (Dunn, 2007), and to promote their engagement in emotional learning and theory of mind development as building blocks for the understanding and use of peer relating skills. Sensory processing intervention in mental health spaces particularly is often overlooked in preference of psychological theories, possibly hindering progress of well-being required for successful and satisfactory experiences.

When working with children and adolescents in these vulnerable populations, it is crucial to look beyond standardised assessments and to link patterns of sensory preferences to typical daily routines, reactions, behaviours and people. Sensory regulation, as with energy levels, naturally fluctuates across an individual's daily routine as they ebb and flow through high- and low-energy zones (Diagram 2). Typically developing children are generally able to regulate their levels independently by, for example, seeking a cuddle when over-excited, allowing them to remain in a state of steady regulation in which they are able to focus, learn or play. Children experiencing mental health or IDD

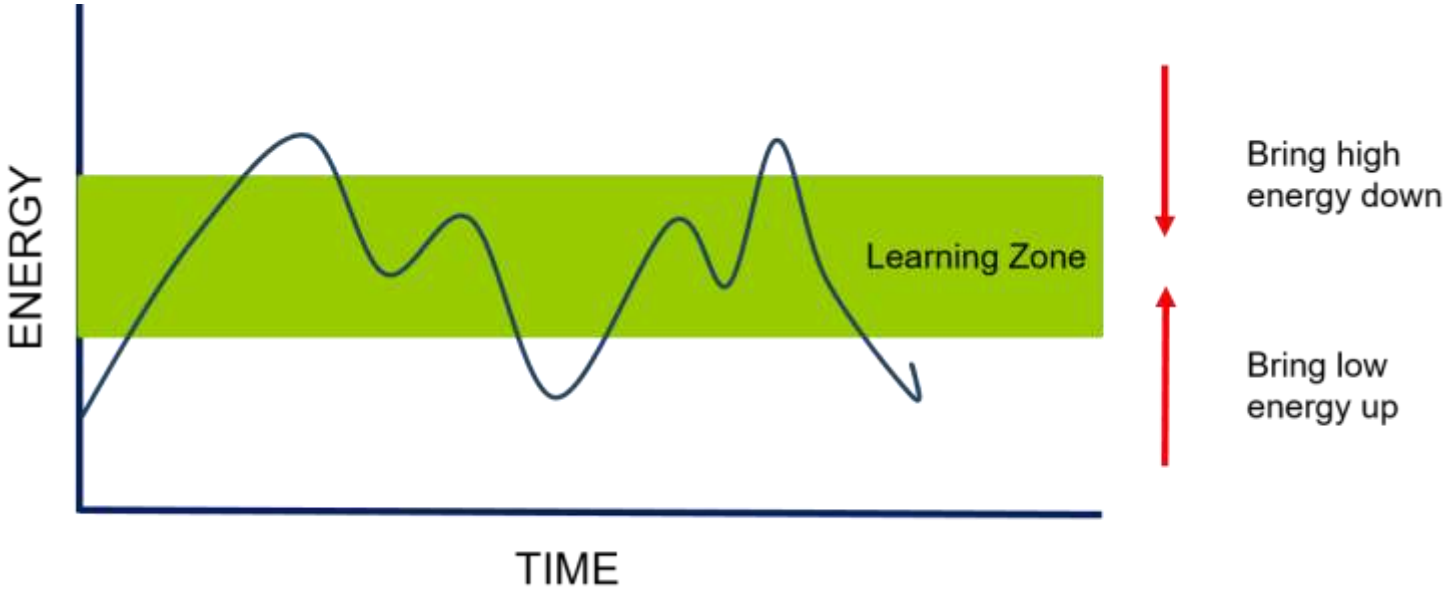


Diagram 2: Example of natural fluctuation of energy across time.

may require additional assistance to recognise their regulation and adjust their energy levels accordingly.

A case study of sensory regulation in practice:
The impact of sensory dysregulation in children experiencing mental health or IDD may be understood by observing a typical daily routine, class or particular behaviour of concern. In a main-stream education class, it is not uncommon for children experiencing mental health and/or IDD to be present without the appropriate adjustments required for them to be functioning in optimal learning zones. A hypothetical case study can be used for examination:

> *This child is 10 years old, living with a diagnosis of Autism Spectrum Disorder with mild learning difficulties. She lives with her mother and two older sisters. She has a history of early childhood trauma in which she witnessed a Domestic Violence relationship between her parents, experienced frequent homelessness and moved house often. Her difficulties in social skills and unregulated behaviour often result in aggressive and deliberate self-harm behaviours. This child is in a mainstream classroom, though is regularly removed from class as a result of her behaviour. A treating team, including occupational therapy and behaviour support therapy, have been employed to assess her daily functioning and provide advice to teacher's and family on targeted behaviour intervention.*

In these cases, standardised sensory assessments may be completed and results used as indicators for intervention planning. However, in vulnerable populations, standardised assessments address only the surface level symptoms. Investigation must be completed across a child's natural routines, environments and relationships in order to determine interventions likely to be most effective. This may be completed by diarising a child's energy levels across a 'typical' day. In our hypothetical case study, this child presents with high levels of energy, held in distress for a large proportion of the day. This increases the amount of time spent outside of the "learning zone", in which a child is able to focus and learn, and decreases the potential for functional gain across all skill domains (see diagram 3).

This must be addressed in an ecological manner in which the information gained from observing the child's sensory preferences and attempts at self- and co-regulate can be used to create proactive sensory plans. These plans include known sensory tasks that work to alert a child who is functioning below the learning zone, to calm a child who is remaining in a high energy zone, and which may be compiled into a

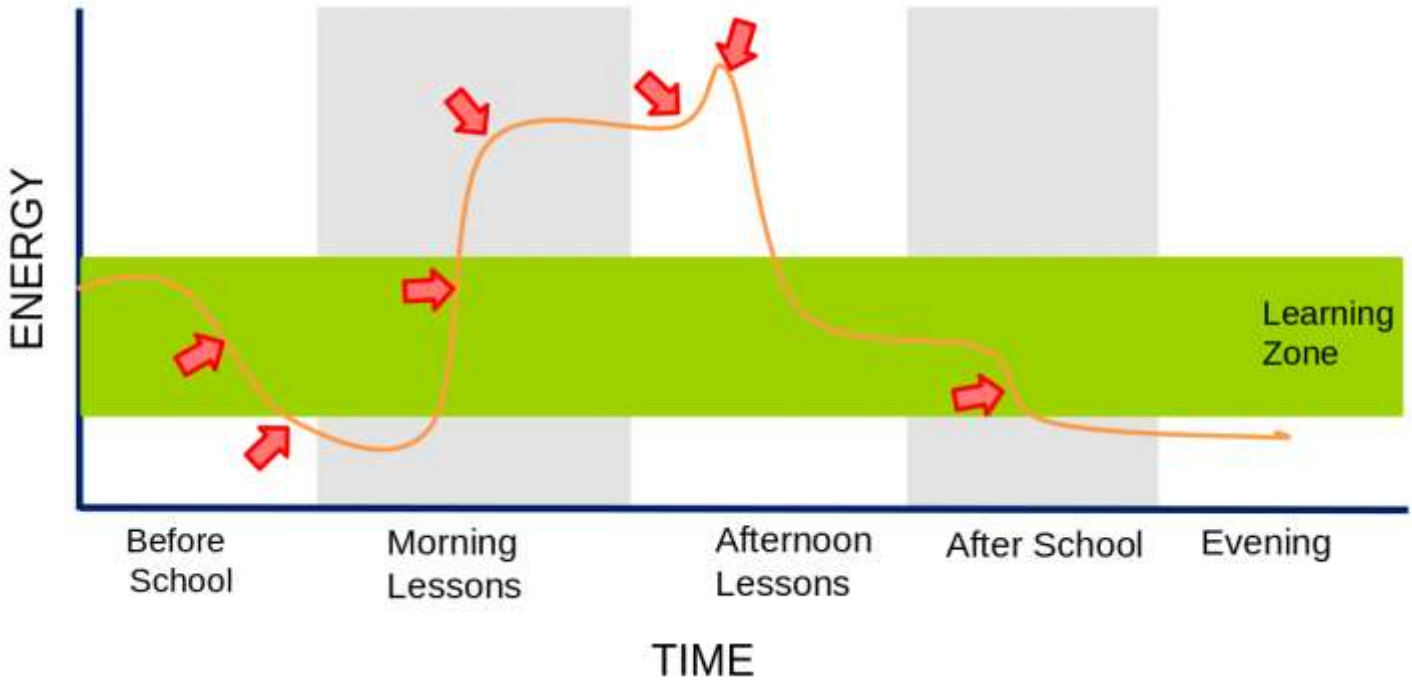


Diagram 3: Example of altered daily fluctuation.

succinct plan for use in home and school environments.

For our case study child, this targeted approach may include:

- *Assistance to heighten energy levels prior to school starting by providing strategies that are alerting to the specific child, such a cold drink through a silicone straw or increasing environmental input by walking barefoot.*
- *Consideration of sensory preferences during classroom tasks to ensure regulation is maintained as long as possible. This may include the use of noise cancelling headphones or dim lighting.*
- *Assistance to de-escalate if heightened energy does occur, such as calm corners, weighted lap toys or music*
- *Assisted bed-time routines including warm showers, warm drinks or deep breathing.*

It should be emphasised that approaches to sensory intervention must be individualised, particularly for children experiencing altered sensory processing. Research into the application of sensory intervention is growing rapidly and should be monitored for best-practice.

References

Camarata, S., Miller, L. J., & Wallace, M. T. (2020). Evaluating Sensory Integration/Sensory Processing Treatment: Issues and Analysis. *Frontiers in Integrative Neuroscience*, *14*. https://doi.org/10.3389/fnint.2020.556660

Child Welfare Information Gateway. (2015). *Understanding the Effects of Maltreatment on Brain Development WHAT'S INSIDE*. https://www.childwelfare.gov/pubPDFs/brain_development.pdf

Dunn, W. (2007). Supporting Children to Participate Successfully in Everyday Life by Using Sensory Processing Knowledge. *Infants & Young Children*, *20*(2), 84–101. https://doi.org/10.1097/01.iyc.0000264477.05076.5d https://depts.washington.edu/isei/iyc/20.2_dunn.pdf

Lyons, S., Whyte, K., Stephens, R., & Townsend, H. (2020). *Developmental Trauma Close Up*. https://beaconhouse.org.uk/wp-content/uploads/2020/02/Developmental-Trauma-Close-Up-Revised-Jan-2020.pdf

Marco, E. J., Hinkley, L. B. N., Hill, S. S., & Nagarajan, S. S. (2011). Sensory Processing in Autism: A Review of Neurophysiologic Findings. *Pediatric Research*, *69*(5 Part 2), 48R54R. https://doi.org/10.1203/pdr.0b013e3182130c54 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3086654/

The Medicine Cabinet: Stopping Medications

Judy Longworth
Clinical Pharmacist
Department of Psychological Medicine
The Children's Hospital at Westmead

When one starts medications, one is often counselled about not suddenly stopping medications and to manage the ceasing of medication under medical advice but why is this so? This does not mean that the medication involved is necessarily addictive as other effects happen in the body as well that I will describe in this article.

Medication can have many different effects on the body and some medications take a long time to have an effect at the therapeutic dose needed; this is because there are receptors involved in different parts of the body and these take time to adjust to being switched off due to the effect of medication or upregulated due to medication having an effect on the pathway that affects this particular receptor. When one stops medication suddenly there can be a build-up of other agents that can have detrimental effects on the body.

For example, when antiepileptic medications are ceased suddenly there can be rebound effects such as epileptic seizures. One might have been seizure free for several years and taking the epileptic medication has a cognitive dulling effect and one just wants it to stop but ceasing suddenly can cause an epileptic seizure and then these can be harder to control. This is also true for some other psychotropic medications. A recent inpatient decided to stop taking an antidepressant by refusing to take and then developed nausea and generalised malaise as well as flu like symptoms. These symptoms ceased when the antidepressant was reintroduced to be weaned appropriately.

Sometimes inappropriate medications are used which may cause more harm than good. This could be because of increased risk of adverse events and additionally, financial costs. Evidence to date indicates the act of ceasing a medication can be as complicated as initiating treatment and the term 'deprescribing' has been used within the geriatric literature for this complex process. There is also a proposed clinical benefit in ceasing use of medications especially when they fall into the same class or similar mechanism of action. Sometimes deprescribing can be used to cease medications that have been added to treat an adverse effect of another medication that is deemed necessary but could have been ceased in the first place such as when benzatropine is added to an antipsychotic when the patient is exhibiting some dystonic reactions but later the antipsychotic is ceased but the benzatropine continues.

Harm associated with ceasing medications include adverse drug withdrawal reactions, pharmacokinetic (what the body does to the drug) or pharmacodynamics (what the drug does to the body) changes and also, the return of a medical condition. These effects can be observed with proper planning and monitoring and even the re-initiation of the medication or equivalent if the condition returns. Examples of pharmacodynamic effects when one ceases fluoxetine or fluvoxamine, both of which have effects on the metabolising enzymes in the body and can affect the blood levels of other medications, can include either excess of the other medication or return of the condition originally being treated. It can take up to one month for the metabolising enzymes to return to base level before the introduction of fluoxetine or fluvoxamine.

Thus, one might not associate the ceasing of the fluoxetine or fluvoxamine with the return of the condition or the need to increase the drug associated with the drug to drug interaction. Return of a condition can also occur when one ceases antiepileptic drugs used to manage both behaviour and epileptic seizures. Usually antiepileptic medications are ceased after a period of time during which the person taking the medication has been seizure free. But antiepileptic drugs can also be used for mood stabilisation and emotion regulation and [although used for this may also for these indications in an epileptic person] and as such ceasing without medical advice might lead to return of the epileptic symptoms or less behaviour control.

“The act of ceasing a medication can be as complicated as initiating treatment...”

Clonidine (See my previous article in Volume 4, Issue 3/4)

When taken to help with arousal and sleep over a long period of time will still have an effect on the blood pressure such that when stopped suddenly there can be a withdrawal reaction causing a significant increase in blood pressure. This can be alleviated if the clonidine dosing is slowly decreased thus the body adjusts over time and thus the rebound hypertension does not occur.

Antidepressants

This group of medications can have significant reported withdrawal effects and can vary between different patients as well as different medications due to their own characteristics and how they affect the body. Selective Serotonin Reuptake Inhibitors (SSRI) include fluoxetine, fluvoxamine, sertraline and (es)citalopram have revolutionised the treatment of depression and other conditions where it is hypothesised a low level of serotonin in the body. The withdrawal reactions listed below are part of complex of symptoms that potentially can occur when one ceases abruptly the SSRI. These reactions have been reported during clinical trial withdrawal as well as the length of time one has been taking the medication might also contribute to the severity of the symptoms. More importantly, some of the symptoms of withdrawal are very similar to the many original symptoms of depression or anxiety that was the reason for the prescribing in the first place. The Serotonin Noradrenaline Reuptake Inhibitors such as venlafaxine and duloxetine have also been reported as having similar symptoms when suddenly withdrawn or ceased.

- dizziness or vertigo
- electric shock sensations in head
- flu-like symptoms
- problems with movement, such as problems with balance or walking, or involuntary movements
- sensory disturbance, such as smelling something that isn't there
- stomach cramps
- strange dreams
- tinnitus (ringing in the ears).

Symptoms like the original problem

- anxiety
- crying spells
- depersonalisation (feeling detached from your surroundings)
- depression
- disturbed sleep
- fatigue (feeling very weary)
- mania
- mood swings
- poor concentration and memory
- suicidal thoughts.

Another group of antidepressants are the tricyclics such as amitriptyline withdrawal reported effects. Although not used as antidepressant specifically amitriptyline is often used to help with sleep and impulsivity when the psychostimulants are not sufficient to man-

age some behaviours especially in the autistic spectrum population.

Can switching antidepressants help with withdrawal?

If you've been taking a drug with a short half-life (time in the body to reduce to half the level), you may experience problems with withdrawal symptoms. In this case, it might be possible for you to switch to a similar drug, but with a longer half-life. You may find this drug easier to come off. For example, this may be switching from an SSRI with a short half-life to another SSRI with a longer half-life. Both paroxetine and venlafaxine have reported withdrawal reactions, so much so that originally in Australia venlafaxine was marketed as an immediate release dosage form but then several years later that form was removed from the market and venlafaxine XR was only marketed in Australia. There is a controlled release form of paroxetine but this form is not available in Australia.

Benzodiazepines such diazepam, lorazepam, alprazolam and temazepam, all have varying half-lives and thus these are subject to psychological as well as physiological dependence and thus the withdrawal needs to be tightly controlled.

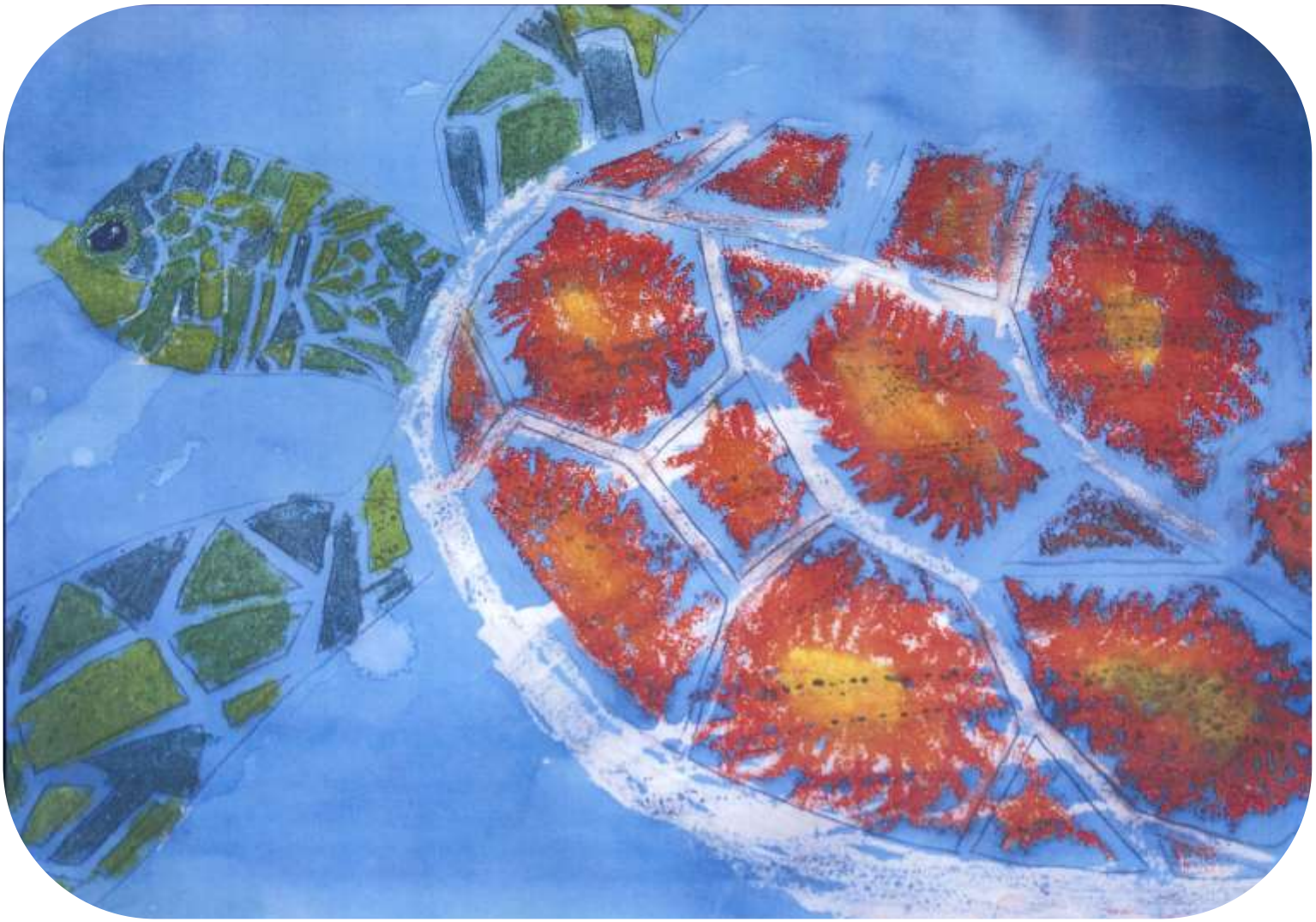
Those which are all long acting benzodiazepines and high potency and thus will take a long time for the body to be clear of any trace. Tolerance to the effects of benzodiazepines are more likely with those used for sedation or epilepsy[4]. Benzodiazepine withdrawal due to sudden cessation can be quite painful and can often mimic the underlying anxiety.

Common withdrawal reactions include these symptoms:

- Nausea
- appetite loss
- irritability
- restlessness
- trouble sleeping
- muscle aches
- tiredness
- anxiety.

These symptoms do not always occur or last long periods of time so under medical guidance these can be monitored and the process done safely.

Although these symptoms are widely documented the frequency of the occurrence has not been documented



and thus needs further studies. The one's individual genetic makeup of the drug metabolising enzymes will have an impact on how one responds to a sudden or gradual withdrawal of antidepressants as well as how long has the course been and the dose.

Risperidone and other serotonin /dopamine antagonists

The process of stopping antipsychotics may be linked to relapse through neuroadaptation that persist after stopping and this includes dopaminergic hypersensitivity[3] .

When ceasing risperidone, weaning needs to occur at the same pace as the initiation of the risperidone. Rebound dyskinesia (erratic involuntary movements) has been reported on withdrawal and this can happen also with inconsistent dosing and sudden ceasing due to adverse effects[1].

Antibiotics[5]

Current thinking about antibiotics is In general, use the shortest possible duration of therapy, consistent with the condition being treated and the patient's clinical response. Prolonged duration of antimicrobial therapy is associated with an increased risk of adverse reactions, *Clostridium difficile* infection, candidiasis and selection of antibiotic-resistant organisms, as well as increased costs.

Examples of indications for which shorter course therapy (less than 7 days) is often appropriate (Box 2.36)

- intra-abdominal infections when definitive surgical management has been undertaken
- uncomplicated lower urinary tract infections
- acute biliary infections when obstruction has been removed
- acute bacterial rhinosinusitis
- acute exacerbations of chronic obstructive pulmonary disease
- community-acquired pneumonia
- uncomplicated skin and soft tissue infections

Other medications

Withdrawal from psychostimulants, both illicit and prescribed, is well established with perceived thinking it was all psychological. In some cases psychostimulants are ceased therapeutically resulting in re-emergence of the ADHD symptoms they were treating(6) The symptoms of withdrawal are unlike the withdrawal symptoms from other illicit drugs such as opioids and the psychostimulant withdrawal syndrome actually mimic those of intoxication, particularly agitation and hyper-arousal.

Nicotine and Illicit drugs are also subject to withdrawal reactions and these can come in the form of cravings but that is for another article.

References:

Sharma A, Shaw SR. Efficacy of risperidone for managing maladaptive behaviours for autistic spectrum disorder: a meta-analysis J Pediatr Health Care 2012; 26:291-9

https://www1.health.gov.au/internet/publications/publishing.nsf/Content/drugtreat-pubs-modpsy-toc~drugtreat-pubs-modpsy-3~drugtreat-pubs-modpsy-3-7~drugtreat-pubs-modpsy-3-7-pws accessed 20/12/21

Horowitz MA, Jauhar S, Natesan S, Murray RM and Taylor D. A method for tapering antipsychotic treatment that may minimize the risk of relapse. Schizophrenia Bulletin 2021; 47: 1116-29

Baldwin DS, Aitchison K, Bateson A, Curran HV, Davies S, Leonard B et al Benzodiazepines: risks and benefits. A reconsideration J Psychopharmacol 2013; 11:967-71

www.tg.org.au.acs.hcn.com.au Antibiotics accessed 25/11/21

Lohr WD, Wanta WA, Baker M, Grudnikoff E, Morgan W, Chhadra D and Lee T. Intentional discontinuation of psychostimulants used to treat ADHD in youth: a review and analysis. Front Psych 12:642798. doi: 10.3389/fpsyt.2021.642798

Useful webpage
https://childmind.org/article/how-to-take-kids-off-medication/

and
https://www.choiceandmedication.org/sydneylocal/printable-leaflets/

Notes from the Conference of the SSBP (Society for the Scientific Study of Behavioural Phenotypes): 8-10th September 2022 in Oslo, Norway.

Dr Peter Wurth, Psychiatrist with interest in intellectual disability.

This face-to-face conference was a welcome reminder of the value of the networking one gets from smaller interdisciplinary meetings. There were also some memorable presentations, plus valuable information about rarer conditions that characterise behavioural phenotypes. Oslo was also an old, pedestrianised city, with trams but Norway has a plethora of the most wonderful, deep, glacial created natural harbours. Edvard Munch is their national treasure artist. We had a mayoral reception at the muralled town hall where the annual Nobel Peace Prize is awarded. Fish soup, giant crabs and reindeer are their local specialties. The fjords are incomparable.

Connie Kasari from UCLA described methodology for improving social communication outcomes for young children with autism and other neurodevelopmental disorders (NDDs). She noted that in the 1980s 70% of children with autism at school entry were minimally verbal compared to 30% today. She claimed this as evidence of the success of early intervention, but it is also evidence of changing diagnostic criteria. Most interventions remain untested but are still very popular. There is generally a poor evidence base, with most studies excluding those with a low IQ, a syndromal diagnosis, low income, low education or complex needs. She promoted the **JASPER** approach of focusing on Joint Attention, Symbolic Play, Engagement and Regulation (www.jaspertraining.org).

Little information is available on those who respond poorly to intervention. 69% of the treatment effect is mediated by the child's capacity for joint attention. Most children with autism come to professional attention because of language delay, even though this is now excluded from the diagnostic criteria.

Her videos showed how a child tends to detach if an adult imposes a framework above their skills but responds to mirroring of their engagement with objects or persons. JASPER combined with an Augmentative Communication Device produces much better outcomes than just using the device. Fears that such devices would reduce the inclination of the child to speak have not been supported. The presence of an individual aide in the classroom is helpful for disruptive behaviours but does not improve social outcomes. Peer mediated interventions to promote inclusion are much more successful than adult mediated approaches.

Kaja Selmer from Oslo University Hospital described the interplay between **epilepsy and behavioural phenotypes**. She noted that the definition of epilepsy is of two unprovoked seizures more than 24 hours apart, but noted that it also includes cognitive, psychological and social consequences. The prevalence is 0.65%, with 8% having autism, 12% having ADHD (attention deficit hyperactivity disorder), 17% having an ID (intellectual disability), and perhaps 80% having some form of behavioural disorder. 5% of epilepsy cases are monogenic. Pathophysiological mechanisms include the presence of an anatomical lesion, disrupted synap-

The Fjords of Norway

The Opera House

tic stability, disrupted networks, neuronal hyperexcitability and metabolic abnormalities.
Tuberous Sclerosis Complex (TSC) is a monogenic, dominant disorder. 80-90% have epilepsy, and 30-50% autism. Treatment with mTOR inhibitors causes a much bigger impact than would be expected by shrinkage of tubers alone. Seizures are reduced, but there is inadequate data to confirm whether symptoms of autism improve. There is a high burden of side effects. Petrus de Vries noted that the average age of the diagnosis of autism in TSC is seven years despite a 50% prevalence, and they therefore miss out on early intervention (yet another case of diagnostic overshadowing).

Juvenile Myoclonic Epilepsy is a multifactorial disorder with unclear genetics. It is the most common epilepsy in adolescence and includes absence and tonic-clonic seizures, but the hallmark is myoclonic seizures. Individuals with this condition have historically been regarded as hedonistic, untrustworthy and immature. There is impaired executive functioning across all domains. Seizures respond well to anticonvulsants, but other problems do not. There is disturbed neuronal connectivity especially in the frontal lobes. There are increased rates of crime, illicit drug use, early smoking, ADHD, troubled parents and being a victim of violence. There is therefore a JME behavioural phenotype. Effectively treatment of myoclonic jerks in this condition reduces impulsivity. Control of these fits should therefore be a priority, whereas it is often regarded as less of a problem compared to controlling major seizures.

Jente Verbesselt from Leuven described behavioural and socio-communicative capacities in children with **16p11.2 deletion** and their syndromes. She noted that this syndrome occurred de novo in 70% of cases, and was associated with an average IQ of 82, motor delay, speech motor problems, autism, obesity, and epilepsy in 20%. Of the 24 individuals studied 10 had autism. Motor delays were the most common reason for genetic testing. The high prevalence of impaired communication is consistent with the high frequency of an autism spectrum diagnosis.

Sissel Helverschou from Oslo University Hospital described the establishment of a **nationwide multicentre mental health service for adults with autism and intellectual disability**. Eight clinical centres were established across the country with a standardised approach to assessment and service delivery. Norway is sparsely populated with large distances. Mental health and intellectual disability services have little overlap. (It is remarkable that eight centres were established in this country with a population of 5.5 million, less than

The Town Hall of Oslo, Norway

that of NSW) They have assessed 132 patients, 89 of whom were male, 87 of whom had a mild or moderate ID and 45 a severe or profound ID. All had autism. Comprehensive assessments one and two years apart showed significant improvements in psychosis, depression and anxiety but not in compulsive behaviours.

Ann Swillen from Leuven described the neurodevelopmental profile and stages of regression in **Phelan McDermid syndrome, a deletion at 22q13.3**. This causes ID, severe speech impairment and autism. It is caused by a deletion that mostly includes the SHANK3 gene. Unknown factors modify the severity of the syndrome. The SHANK3 gene is associated with ID, autism, epilepsy, ADHD and bipolar disorder. There is a coarse face. More than 25% have epilepsy and more than 50% have autism. They have a high pain threshold, gastro-oesophageal-reflux, they overheat and turn red easily and have reduced sweating. There are neonatal hypotonia and delayed motor milestones.

She identified four stages of regression in 24 patients

followed long-term. 79% had shown a significant loss of skills over the lifespan. Active vocabulary was the main loss followed by psychosocial adaptability, fine motor skills and walking. The first stage was an abrupt decline in speech, the second was a prolonged period of stagnation rather than continuing development. The third showed an acute neuropsychiatric decline often with catatonia, hallucinations and psychosis. The fourth and final stage was often precipitated by an event such as a severe illness or psychosocial stress, and was characterised by profound motor regression sometimes to the point of wheelchair dependence. In some the decline is so dramatic that the patient is taken to the emergency department because of suspicions of acute illness. Regression is a key feature of this syndrome, with the first stage occurring at around seven years, followed by a long period of stagnation, with the third stage at around the age of 20 and the final stage at 27. The mechanism is unknown. Some patients with catatonia responded to lithium (not a recognised treatment for other causes of catatonia). Peter Wurth commented anecdotally on regression in Downs responding to IV gamma globulin.

Lauren Shelley from Aston University, UK presented on **aggressive behaviour in SATB2-Associated Syndrome**. This syndrome was described in 2014 and has also been called Glass Syndrome. It is estimated to explain 0.25% of undiagnosed ID. The SATB2 gene is located at 2q33.1. It causes a moderate to profound level of intellectual disability, the absence of speech, osteoporosis and dental and palatal problems. It is associated with a happy, hypersocial disposition. There has been an estimated prevalence of 77% for aggressive behaviour. 37 caregivers of individuals whose age ranged from 3 to 33 completed questionnaires and interviews. 86% of the patients had engaged in aggressive behaviours in the previous months.

Honey Heussler from Brisbane presented an open label study on the efficacy of transdermal **synthetic cannabidiol gel ZYN002 for the treatment of young people with 22q11.2 deletion syndrome** (DiGeorge/VCFS). This is a pharmaceutically rather than plant derived form of cannabidiol under investigation for the treatment of behavioural symptoms in the above syndrome, FXS (Fragile X Syndrome) and autism. 20 patients, mean age 9.9 years, with 60% male were enrolled. There were improvements in global anxiety, irritability and hyperactivity, with minimal, transient side effects. Patients with a history of cardiac disease were excluded, which caused difficulty in recruiting given the prevalence of cardiac disease in this syndrome. This product does not undergo gastric conversion to THC (tetrahydrocannabidiol) unlike plant derived cannabidiol products. These preliminary results support the need for formal RCTs which are underway.

Thomas Werge from Copenhagen presented on the need to move from the traditional **cross-sectional approach to a longitudinal approach** to diagnosis and noted the clustering of genetic vulnerability to various groups of diseases. Trajectories were constructed for Danish individuals with schizophrenia showing vulnerabilities to various comorbidities. For example mental health diagnoses seem to correlate with pulmonary diagnoses (eg asthma) as well as neurological. But genetic risk to schizophrenia is associated with creativity.

Ida Sonderby from Oslo University Hospital presented on the use of carriers of recurrent **rare Copy Number Variants (CNV) to gain insight into neurodevelopment and disease**. She noted that reciprocal CNVs at one locus, i.e. deletions and duplications, could be associated with the same disorder. Additionally, one specific CNV may increase the risk for several different disorders. A range of 7 CNVs all create a high risk of ADHD, autism, ID, epilepsy and schizophrenia, and of a range of somatic diseases including obesity, renal failure, cataracts, immunological disease and an increased risk of early death. These risks are present even in those without an early NDD. For example the 22q11.2 deletion and duplication both cause intellectual disa-

bility, autism, ADHD and possibly schizophrenia. Brain structure is very heritable, with many more changes in monozygous compared to dizygous twins but notably there is discordance even between monozygous twins.

The ENIGMA study in Norway has recruited a cohort of 140,000 children and parents, with most genotypes, and many having had brain and body MRIs. The cohort is now in their 20s and 93 CNVs have been investigated. 4,500 have ADHD, of whom 220 have one of seven CNVs. Each CNV has specific effects on brain structures which may be independent of the phenotype.

Ellen Langballe presented the **Norwegian Down syndrome and dementia study**. She noted that in 1993 the life expectancy of an individual with Down syndrome was 25, in 1997 49 and in 2018 60. 90% will develop dementia, and the frequency is increasing as the lifespan increases. The age range of onset is 35-74 with the average 55. 50% are within the sixth decade, and survival post-diagnosis averages 3.5 years. Those with ID without Down syndrome show an increased dementia risk starting from 50, but with a much lower prevalence. Trisomy 21 causes an overdose of 300 genes, one of which is the amyloid precursor protein gene. A large study following individuals with Down syndrome to investigate the usefulness of psychometric tools and plasma biomarkers was due to start in March 2020 but postponed by the COVID epidemic. This survey is underway now with no results yet available.

Vigeband Sculpture Garden

Jeanne Wolstencroft from Great Ormand Street Hospital/UCL London presented the **IMAGINE-ID longitudinal study of mental health in UK children with ID of known genetic aetiology**. Substantially higher rates of mental illness and behavioural difficulties were found between these children and neurotypical children, with more than 50% in the very high severity range on the Strength and Difficulties Questionnaire compared to 5% of UK norms. 17% of these ID children were in the average range on the questionnaire compared to 80% in the UK norm population.

Sabine Mouse from Rotterdam presented on **autism symptom profiles in young people with FXS, NF1 (Neurofibromatosis 1), TSC and Angelman syndrome**. She found large variabilities in the autism symptoms exhibited by individuals with these different symptoms and noted the requirement for a personalised approach.

Ramkumar Aishworiya from Singapore presented work completed at UC Davis on the clinical **phenotype and molecular biomarkers in FXS**. She noted that FMR1 is now renamed the Fragile X Messenger Ribonucleoprotein rather than the Fragile X Mental Retardation Protein. FMRP has multiple actions including suppressing MMP9 and the autism risk gene CYF1P. She noted that elevated levels of MMP9 have been associated with increased weight in FXS, and an audience member noted that these elevated levels are also seen in Prader Willi syndrome. Elevations in MMP9 and the CYF1P gene will be used as biomarkers to track the efficacy of a metformin RCT in FXS.

Elise Pelgrims from Leuven described the phenotype of terminal triplications of 1p36.3. Her team has added two more cases to the one patient previously described with this rare triplication, all of which show distinct facial dysmorphia together with severe ID, hypotonia, epilepsy and cardiovascular diseases. Prominent dysmorphic features include hypertelorism and ptosis. Many of these features are shared with 1p36 duplications. The 1p36 deletion syndrome is seen in 1/ 5000 births and features ID, motor and speech delays, epilepsy, hypotonia and cardiovascular disease.

Cristan Farmer from NIMH USA described a method for demonstrating clinically meaningful **improvement in cognitive and adaptive behaviour ability scores**, crucial for demonstrating the efficacy of interventions re-

quired to obtain funding. The challenge with interventions in children with various NDDs is that they are developing and therefore changing, presenting a moving target for assessment and for the demonstration of intervention efficacy. The probability of the individual achieving developmental milestones may be more useful that referencing change against population norms.

Charlotte von der Lippe from Telemark Hospital, Norway presented on the **why, what, when and how of genetic testing**. She noted that adults with ID get much less attention for genetic investigation and response to findings than children. The why is to establish whether there is cause, whether there is anything treatable, what the likely course of illness is, what screening for comorbidities is required, what the risk to siblings is, for the purpose for research, to enable families to contact peer groups, and with positive findings, often to relieve maternal guilt.

What tests are done is determined by whether biological parents are available. A Comparative Genomic Hybridization (CGH, a method for analyzing CNVs) and microarray plus Fragile X DNA are basic, with whole exome sequencing and trio testing focusing on areas of concern identified in the patient, and testing these areas in the parents. Normal individuals have 200 rare variants on WES (whole exome sequencing). This testing reveals whether the stated parents are the biological parents, and parents must be warned about this. The exome is 1% of the genome but is responsible for 80% of gene disorders. Doing a whole genome sequence is much more work.

Newborns are often tested for conditions that are treatable, and that have consequences without treatment. In Norway 26 diseases are screened in newborns. A major question is whether parents are ready. Testing can be both diagnostic and predictive (e.g. for carrier status in cystic fibrosis).

The how it is done – blood samples are better but buccal smears can suffice. Parents often have a lot of fears of stigma, shame, insurance problems and unrealistic hopes. Surprisingly many are motivated by desire to help others. It is important to note that genetic counselling is voluntary and it cannot be assumed that families want it.

Donna McDonald-McGinn from Philadelphia presented findings in 22q11.2 CNVs diagnosed in adulthood. She noted that on theoretical grounds duplications should be as frequent as deletions. The deletion syndrome occurs in 1/2000 births and is the most common microdeletion. IQ is typically around 70, and problems include renal agenesis, hypocalcaemia, epilepsy, ADHD, anxiety, autism, and schizophrenia in 25%. The phenotype in monozygous twins varies. The average age at diagnosis is 3.9 years. It is often first diagnosed in adulthood if there is no CHD. IQ can range from severe ID to average.

Duplications are mostly familial with an average IQ of 84. Schizophrenia is seen despite early reports suggesting that this duplication might be protective. Also common are ADHD, anxiety, autism and epilepsy. Congenital heart disease occurs in 18% compared to 37% in the deletion. There are palatal and renal abnormalities.

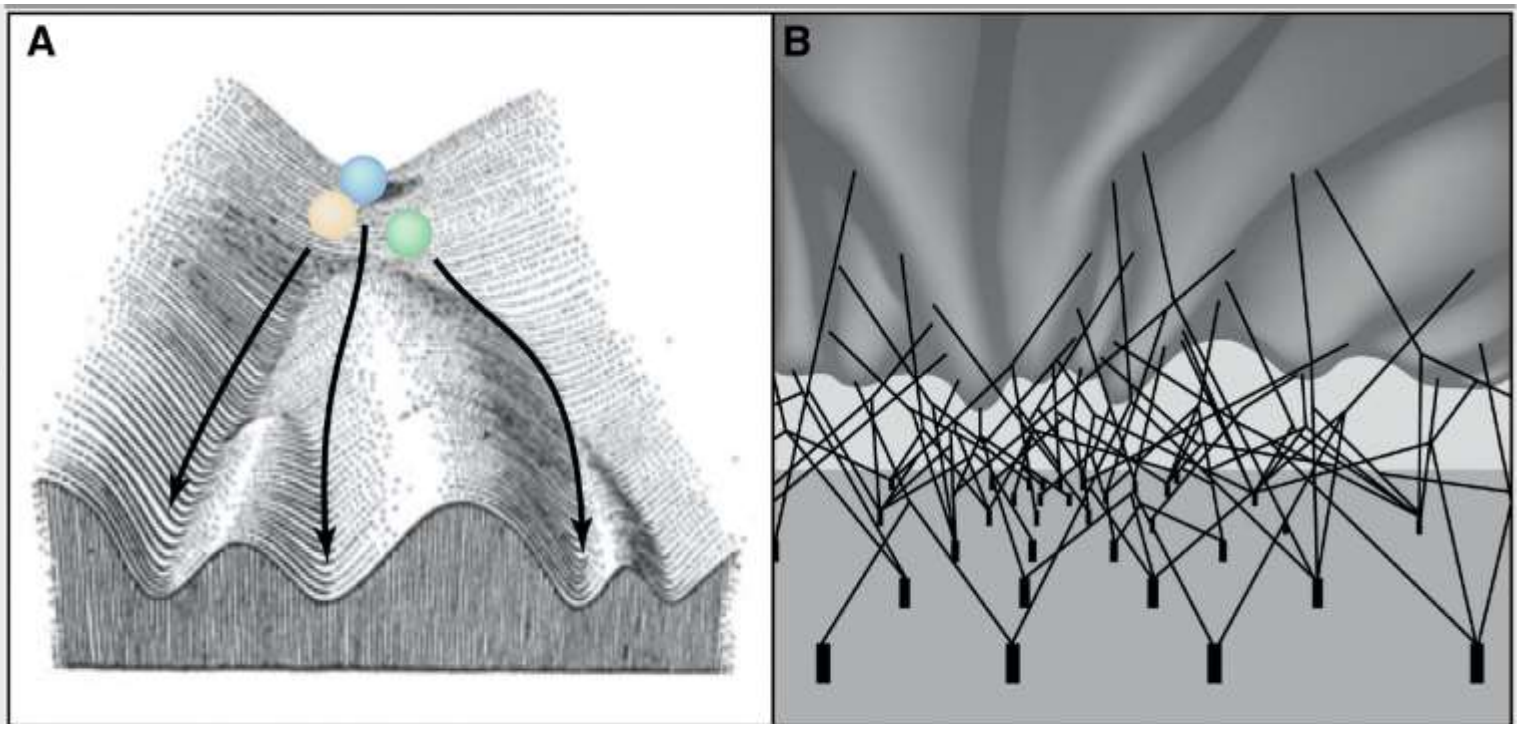
Nicole Tartaglia from University of Colorado presented on the developmental profiles of prenatally diagnosed sex aneuploides especially trisomies. Most of these individuals have been diagnosed postnatally, and the phenotype is significantly more severe than in those diagnosed prenatally who might otherwise be missed. Conditions include XYY and XXX, both seen in 1/1000 births, XXY or Klinefelter's seen in 1 in 650 births, XO or Turner's syndrome seen in 1/2500 births, with the tetrasomies XXYY and XXXY also identified. Learning difficulties, ADHD and anxiety are common in these conditions. Since 2011 non-invasive foetal DNA test-

ing has been able to be carried out on maternal blood. One in 400 babies has one of these conditions. There is however a potential negative consequence of diagnosing these individuals prenatally in terms of labelling and expectations for a greater number of problems than may in fact naturally occur. On the other hand early treatment especially for Klinefelter's can improve development. It is important to remember that genetics is not destiny.

Kevin Mitchell Behavioral Geneticist from Trinity College, Dublin outlined **'What we have learned from psychiatric genetics'** (Kevin J Mitchell, 2018. Innate*:* How the Wiring of Our Brains Shapes Who We Are*.* Princetown University Press). This was the highlight of the conference. His central theme was that many phenotypes in psychiatry are emergent depending on multiple factors. Non-genetic is not the equivalent of environmental aetiology. Development is probabilistic and the process is 'noisy'. Genes encode biochemical rules, not outcomes. Every time the program (genotype) is read the outcome (phenotype) varies. The brains of monozygous twins show differences at birth, with the potential for cascading processes causing greater divergence. He showed Waddington's epigenetic landscape diagram from the 1940s, and a refinement of this diagram showing the valleys and hills of this diagram being caused by underlying factors including the role of multiple genes. There are risk genes rather than disease genes, and a given risk gene can for example increase the chance of autism, ADHD, schizophrenia and bipolar disorder. He introduced the concept of developmental robustness to describe the integrity of the genome and the extent to which the phenotype may be altered by a given risk gene. A risk gene may therefore have minimal effect in an individual with developmental robustness, whereas in another individual it may cause significant phenotypical difference.

The central problem with psychiatric genetics is that we see the same phenotype with multiple genetic pathways, and the same gene causing multiple phenotypes. Personalised medicine at a genetic level is therefore an unlikely prospect. There are 300-400 genome wide hits in schizophrenia which create tiny individual effects.

He noted that there are two major classes of neurodevelopmental disorders – the rare, such as FXS, Williams syndrome etc., with simple genetics, and the common including ID, autism, epilepsy and schizophrenia, with complex inheritance. Some conditions are no doubt caused by a combination of rare variants and multiple common variants. This talk therefore was very helpful in explaining why we have such little clear information about the cause of major mental illness. Measures to enhance robustness are the most likely to improve outcomes. Mitchell predicts we are 30 years away from understanding how genetics influence the variance of development of proteins, biochemical functions, cells specialisation, circuits, anatomy and physiology that lead to development of the brain and psychological traits.



Waddington's epigenetic landscape. Illustration of the randomness of developmental variance (1957)

The beautiful artworks in this journal are taken from the participants of the **Operation Art project** at the Children's Hospital at Westmead. You can find out more at https://www.artsunit.nsw.edu.au/visual-arts/operation-art-2014

contact us...

The Children's Hospital at Westmead
School-Link Initiative
Department of Psychological Medicine
Cnr Hawkesbury Rd and Hainsworth St,
Westmead NSW 2145
SCHN-CHW-SchoolLink@health.nsw.gov.au

P: 9845 2005
W: www.schoollink.chw.edu.au

If you would like to contribute to our next edition, please contact;

CHW School-Link journal
MHCAIDD editor
Hebah Saleh
hebah.saleh@health.nsw.gov.au