

“Autism Spectrum Disorder (ASD) and Attention Deficit Hyperactivity Disorder (ADHD): how to go about diagnosing or seeking help if you are concerned your child is affected”

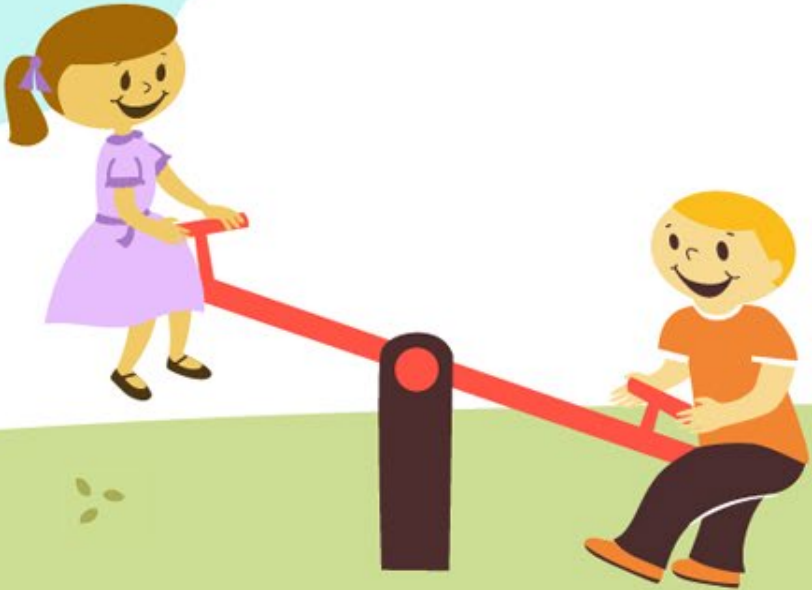


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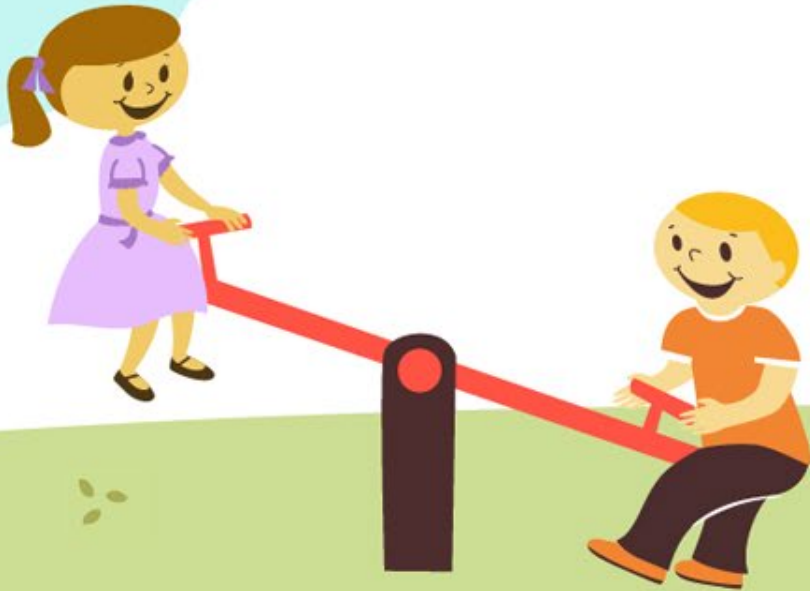


Tuberous Sclerosis Complex (TSC)

- characterized by pleomorphic features involving many organ systems, including multiple benign hamartomas of the brain, eyes, heart, lung, liver, kidney, and skin



- Most patients with TSC have epilepsy, and one-half or more have cognitive deficits and learning disabilities
- Other common manifestations include autism, behavioural problems, and psychosocial difficulties eg, anxiety
- Collectively, these are termed TSC-associated neuropsychiatric disorders (TAND)



- These problems are usually associated with brain lesions including glioneuronal hamartomas (also called tubers), periventricular giant cell astrocytomas, and abnormalities of cerebral white matter detected on neuroimaging studies
- However, there is a wide variety of phenotypes between and within families regarding the number and severity of TSC manifestations



Cognitive deficits

- Cognitive disability/deficit is a primary feature of TSC, affecting 44 to 65 percent of patients (the range of intelligence in affected patients is highly variable)
- It is associated with a history of infantile spasms, refractory seizures and, to a lesser extent, number of glioneuronal hamartomas
- In a cohort of over 1600 patients from a TSC database, epilepsy onset before the age of two years was associated with a higher frequency and severity of intellectual disability
- Epilepsy was also associated with autism and attention-deficit-hyperactivity disorder

Autism spectrum disorder (ASD)

- Neurodevelopmental disorder characterized by persistent deficits in:
 - social communication and social interaction and
 - restricted, repetitive patterns of behaviour, interests, and activities



Autism and behavioural problems

Autism and autistic behaviours, including hyperactivity, inattention (ADHD), and self-injurious behaviour, are common in children with TSC and can be a significant source of stress for parents and caregivers



- In different case series, the prevalence of significant behavioural problems among children with TSC ranges from 40 to 90 percent
- While behavioural problems can occur in the setting of either normal intelligence or cognitive dysfunction, at least one case series found that low intellectual functioning and higher seizure frequency were risk factors for behavioural disorders



- Whether autism is associated with a specific location of glioneuronal hamartomas is **uncertain**
- In one study, development of an autism spectrum disorder was associated with the presence of temporal lobe glioneuronal hamartomas, temporal lobe epileptiform discharges, and early onset of persistent infantile spasms
- However, others have shown that the frequency of glioneuronal hamartomas in the subcortical or cortical regions was similar in TSC patients with and without autism

Early symptoms and signs of autism

Parental concerns about deficits in social skills

Parental concerns about deficits in language skills or behavior

Parental concerns about frequent tantrums or intolerance to change

Delayed language and social/communication skills:

- Lack of orientation to name by age 12 months
- Lack of pointing or gesturing to indicate interest (eg, by pointing to an airplane flying over) by age 14 months
- Lack of pretend play (eg, "feeding" a doll) by age 18 months

Avoiding eye contact or wanting to be alone

Having trouble understanding other people's feelings or talking about their own feelings

Repeating words or phrases over and over (echolalia)

Giving unrelated answers to questions

Getting upset by minor changes

Having obsessive interests

Flapping their hands, rocking their body, or spinning in circles

Having unusual reactions to the way things sound, smell, taste, look, or feel

Phenotypic spectrum and clinical presentation of ASD

- Presentation in the first two years of life with characteristic features as per the table before
- Approximately two-thirds of patients with ASD present with lack of acquisition of communication skills before age two years
- Plateau of social skills after typical early development



Phenotypic spectrum and clinical presentation of ASD

- Approximately one-fourth to one-third of children with ASD achieve early language milestones but have regression or plateau of language, communication, and/or social skills between 15 to 24 months of age
- The regression of skills can be gradual or sudden and may occur in the context of pre-existing developmental delays or atypical development



Phenotypic spectrum and clinical presentation of ASD

- Lack of interest in others, failure in empathy, absent or delayed language and communication, marked resistance to change, and restricted interests during the preschool years
- For children with less severe phenotypes, parents or teachers may notice behaviour disturbances (eg, overfocus on preferred topics, difficulty with organization, disruptive behaviours) or
- Symptoms related to an associated or comorbid condition (eg, anxiety, attention deficit hyperactivity disorder) before atypical social skills (eg, lack of social awareness or social ability) or atypical language skills are apparent



ASD surveillance and screening

ASD surveillance is encompassed in routine developmental-behavioural surveillance and screening, which consists of:

- eliciting parental concerns
- maintaining a developmental history
- observing parent-child interactions
- identifying risk and protective factors
- periodic developmental screening



- Routine developmental surveillance is recommended at every well-child visit with the GP and Child Health nurses and during immunisation visits
- Allied Health therapists also usually keep surveillance and do screenings or refer for assessments if suspicious of red flags for ASD or behavioural difficulties
- TSC clinic at the QLD Children's Hospital has a multidisciplinary team including a developmental paediatrician and neurologist who reviews and monitors



Benefits of surveillance/screening

- Early identification is an important step in optimizing outcome; early identification facilitates:
 - Earlier education planning
 - Family support
 - Delivery of appropriate medical care and treatment of associated conditions
 - Early intensive intervention that is individualized according to the specific strengths, weaknesses, and needs of the child and family. Early and appropriate interventions are critical for optimizing outcomes in individuals with ASD



Diagnosing Autism Spectrum Disorder

- Symptomatic children with clinical features associated with ASD should undergo ASD-specific screening as part of their evaluation. Symptomatic children include:
 - Children with delayed language/communication milestones identified by screening tests or parental report
 - Children with regression in social or language skills
 - Children with a sibling diagnosed with ASD (family hx is important)
 - Children (regardless of age) whose parents, care provider, or clinician raise concerns regarding ASD (eg, those with atypical behaviours, difficulty socializing, rigidity of behaviour that interferes with function)



ASD diagnostic tools

A number of diagnostic tools are available eg

- Autism Diagnostic Observation Schedule-2nd edition (ADOS-2)
- Childhood Autism Rating Scale 2nd edition-CARS-2
- Autism Diagnostic Interview-Revised (ADI-R)



- Diagnostic tools are used to gather behavioural data in a structured and consistent manner, either through an interview with the caregivers or (more commonly) by making direct observations of the child
- Diagnostic tools are used in conjunction with clinical judgment to make a diagnosis of ASD; **they should not be used in isolation**



- The reference standard for evaluating a child for ASD consists of a comprehensive assessment.
- A comprehensive assessment includes a multidisciplinary team with a lead clinician who has expertise in the diagnosis and management of ASD (eg, a developmental-behavioural paediatrician, child psychiatrist, child neurologist, or [neuro]psychologist), a speech and language pathologist, and an occupational therapist.
- Each of the team members may evaluate the child at a single coordinated visit or individually at different visits.

Psychosocial history

- Very important
- Should include information regarding the family supports and stresses, which may affect management
- Exposure to trauma, early deprivation, and attachment disorder (mainly children who are in the care of the department of child safety) can result in symptoms that overlap with ASD but need to be differentiated from ASD because they require treatment separate from the treatment for ASD

Ancillary testing

- Necessary to corroborate and confirm the diagnosis of ASD, to differentiate ASD from other conditions that mimic ASD, and to identify conditions that occur with ASD
- In addition, ancillary testing helps to determine the child's level of impairment and identify targets for therapeutic intervention

Ancillary testing generally includes:

- Speech, language (including pragmatic language), and communication assessment
- Developmental/intelligence testing with separate estimates for verbal and nonverbal skills
- Assessment of adaptive skills
- Sensorimotor and/or occupational therapy evaluation
- Vision and hearing assessment (if not already performed)

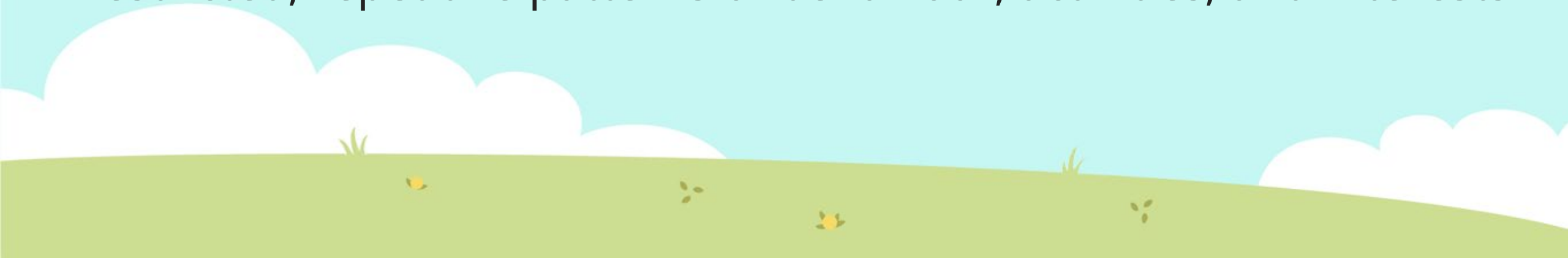
The diagnosis of ASD is made clinically in children who meet established diagnostic criteria for ASD based on history and observation of behaviour

There are two major sets of **diagnostic criteria**

1. Diagnostic and Statistical Manual of Mental Disorders (DSM)
2. International Disease Classification (ICD)

Both of which centre on:

- atypical social communication and interaction and
- restricted, repetitive patterns of behaviour, activities, and interests



Assessment of severity

- In conjunction with an adaptive scale, we use the DSM-5 classification to specify the severity level of ASD, recognizing that severity may vary with context and over time
- **Level 1** ("Requiring support") – Noticeable impairment without support
- **Level 2** ("Requiring substantial support") – Marked deficits in communication; impairments apparent even with supports
- **Level 3** ("Requiring very substantial support") – Severe impairments in functioning



ASD: Overview of management

Goals — The overarching goals of treatment are to maximize functioning, move the child toward independence, and improve the quality of life.

Specific goals address the core deficits of ASD and seek to:

- Improve social functioning and play skills
- Improve communication skills (both functional and spontaneous)
- Improve adaptive skills
- Decrease non-functional or negative behaviours
- Promote academic functioning and cognition



Early intervention

There is a consensus that treatment for ASD must be individualized depending upon the specific strengths, weaknesses, and needs of the child and family

Early diagnosis and early intensive treatment have the potential to affect outcome, particularly with respect to behaviour, functional skills, and communication

Although there is no cure, symptoms can decrease over time and in a small minority be minimized to the extent that they no longer cause disability

There is increasing evidence that intervention is more effective when initiated as early as possible. The establishment of appropriate management strategies in the early years can help to minimize or even avoid subsequent behavioural problems

Specialist involvement

Children with a diagnosis of ASD should have ongoing follow-up with a specialist (eg, developmental and behavioural paediatrician, neurologist, psychologist, psychiatrist)

or a team of providers who can monitor progress, provide recommendations for behavioural programming, and screen for medical concerns



Treatment setting

Therapies for children with ASD may be provided by an early intervention program, school-based special education program, or by therapists in private practice



Treatment for autism spectrum disorder (ASD) focuses on behavioural and educational interventions that target the core symptoms of ASD ie, deficits in social communication/interaction and restricted, repetitive patterns of behaviour, interests and activities

Pharmacologic interventions may be used to address medical or psychiatric comorbidities or provide symptom control but do not treat the core deficits



Specific treatment programs should be individualized according to the child's functioning and needs

Treatment programs should be monitored to ensure appropriate response to therapy

The program should be reviewed and modified as the child's needs change over time



Behavioural and educational interventions

The importance of early intensive behavioural and educational interventions in improving outcomes for children with ASD is well documented

Core features of successful autism educational programs include:

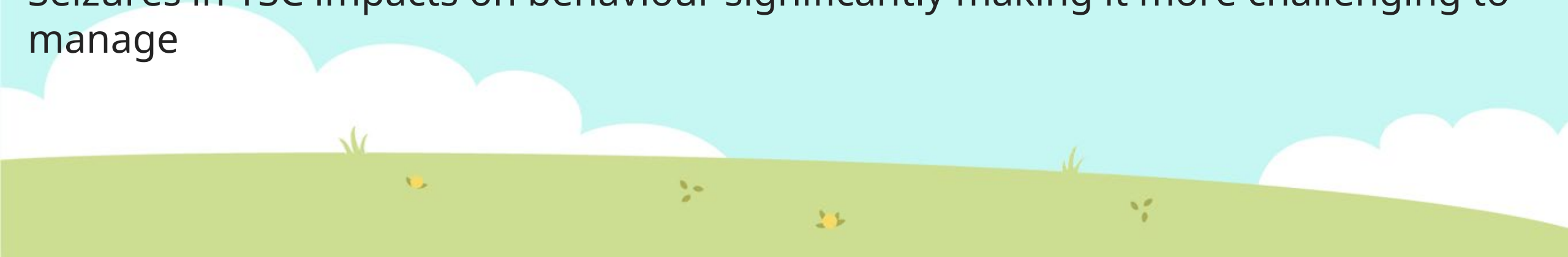
- A high staff-to-student ratio (1:1 or 1:2)
- Individualized programming for each child
- Teachers with special expertise in working with children with autism
- A minimum of 25 hours per week of services
- Ongoing program evaluation and adjustment
- A curriculum emphasizing attention, imitation, communication, play, social interaction, regulation, and self-advocacy
- A highly supportive teaching environment
- Predictability and structure
- Functional analysis of behaviour problems
- Transition planning
- Family involvement
- Close monitoring and modification as the child's needs change

Psychopharmacologic interventions

Psychotropic medications often are used in children with ASD to treat targeted symptoms including:

- Hyperactivity, inattention, and impulsivity
- Aggression, outbursts, and self-injury
- Anxiety
- Obsessive-compulsive behaviours, rigidity, and repetitive behaviours
- Depressive symptoms
- Sleep dysfunction

Seizures in TSC impacts on behaviour significantly making it more challenging to manage



Psychopharmacologic interventions do not treat the underlying ASD. However, they can improve the child's functioning and the ability to participate in behavioural interventions

Psychopharmacologic interventions should be used in conjunction with appropriate behavioural and environmental interventions

Some medications commonly used for behaviour management in ASD:

- Risperidone - for aggression, self injurious behaviour
- Aripiprazole- for aggression, self injurious behaviour (not on PBS)
- Sertraline/ Fluoxetine/ Fluvoxamine- for anxiety, OCD
- Stimulants eg Ritalin/Ritalin LA/ Concerta/Dexamphetamine – ADHD
- Clonidine- ADHD, sleep
- Guanfacine – ADHD
- CBD trial (for anxiety in ASD)

Family support

- Supporting the family is crucial
- Generally family support is via:
 - The primary care provider- GP
 - Specialists
 - Nurse Navigator
 - Complex care teams/Clinical Nurses
 - NDIS
 - Education system/school
 - Allied health therapists



ASD prognosis

- **Core symptoms** — it is difficult to predict outcomes for children with autism spectrum disorder (ASD), especially for children younger than three years
- Some children will retain the diagnosis despite improvement in core symptoms; others, particularly those with milder symptoms, may no longer meet diagnostic criteria for ASD
- Typically, children who no longer meet criteria for ASD demonstrate some residual social, language, and behavioural symptoms and may meet criteria for language, attentional, and/or mood disorders



Factors that have been associated with positive outcomes in ASD children include:

- Presence of joint attention
- Functional play skills
- Higher cognitive abilities
- Decreased severity of ASD symptoms
- Early identification
- Involvement in intervention
- A move toward inclusion with typical peers

Factors that have been associated with less favourable outcomes in ASD children include:

- Lack of joint attention by four years of age
- Lack of functional speech by five years of age
- Intelligence quotient (IQ) <70
- Seizures or other comorbid medical or neurodevelopmental conditions
- Severe ASD symptoms



Social function and employment

- Data for individuals with ASD are limited and mostly based on a few population sample studies. In many of these studies, social functioning continued to be a concern
- One sample of adolescents and young adults with ASD showed, social isolation was common (55 percent had not gotten together with friends in the previous year; 64 percent had not received any phone calls from friends in the previous year)
- Factors associated with more social interaction included the ability to communicate effectively, less severe ASD, lack of poverty, and having parents who advocated for them at school



Although not examined in the context of social functioning, many individuals with ASD found employment over time

Participation in postsecondary education or employment increased with the number of years out of high school (48 percent at one to two years, 89 percent at four or more years) and was associated with higher functional ability and higher parent or guardian household income



Employment did not necessarily equate with independence, which was affected by multiple factors

In another long-term study of adults with varying levels of cognitive function, only 12 percent achieved a high level of independence, and overall outcome was variable, even in those with IQ >70

Relatively few married or were involved in reciprocal relationships, highlighting the persistent deficits in social functioning described above



Attention Deficit Hyperactivity Disorder

- Neurodevelopmental disorder- common in TSC
- Presents with symptoms of hyperactivity, impulsivity, and/or inattention
- The symptoms affect cognitive, academic, behavioural, emotional, and social functioning
- Risk factors are low intellectual functioning and higher seizure frequency
- Prevalence of significant behavioural problems among children with TSC ranges from 40 to 90 percent
- Co-morbid to ASD

Diagnostic criteria for ADHD include symptoms of hyperactivity, impulsivity, and/or inattention that occur in more than one setting and affect function (eg, academic, social, emotional, etc)

DSM 5 criteria is used

Conner's Questionnaire used to assess – Parent and Teacher



Target goals

The management of children with ADHD centers upon the achievement of target outcomes that are realistic, achievable, and measurable

Examples of target outcomes include:

- Improved relationships with parents, teachers, siblings, or peers (eg, plays without fighting at recess)
- Improved academic performance (eg, completes academic assignments)
- Improved rule following (eg, does not talk back to the teacher)

Treatment of coexisting conditions

As many as one-third of children with ADHD have one or more coexisting conditions (eg, learning disabilities, oppositional defiant disorder, conduct disorder, anxiety disorder, mood disorders, tics, sleep disorders)

It is important to treat coexisting conditions concurrently with ADHD

Treatment of coexisting conditions may influence the treatment for ADHD.

Adjunctive behavioural and psychologic interventions may be necessary for managing the full spectrum of symptoms in children with ADHD and coexisting conditions.

ADHD: Overview of management

Preschool children

Behaviour therapy initially

The addition of medication to behaviour therapy may be indicated if target behaviours do not improve with behavioural therapy and the child's function continues to be impaired.

Examples of situations in which medication may be warranted for preschool children include:

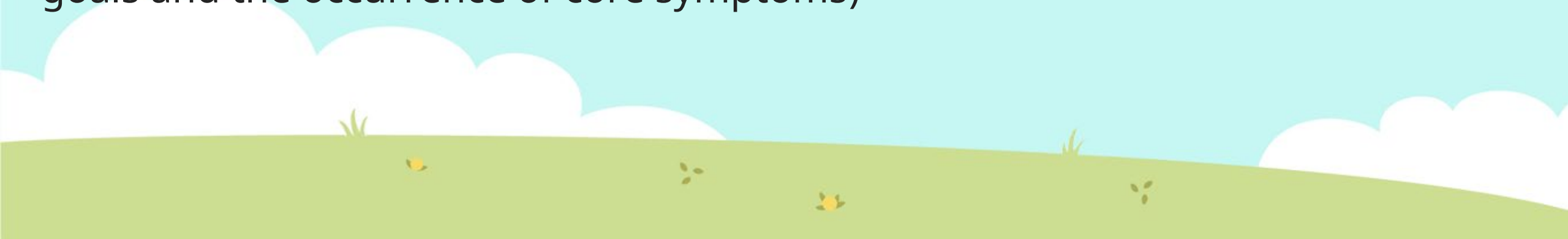
- Expulsion (or threatened expulsion) from preschool or daycare
- Significant risk of injury to other children or caregivers
- Strong family history of ADHD
- Suspected or established central nervous system injury (eg, prenatal alcohol or cocaine exposure)
- ADHD symptoms interfere with other needed therapies

School-age children

For most school-aged children and adolescents (≥ 6 years of age) who meet the diagnostic criteria for ADHD, treatment is behavioural therapy in combination with medication to improve core symptoms and target outcomes

Comorbid conditions must be considered in selecting a specific treatment approach

Monitoring: Children undergoing treatment for ADHD should be monitored regularly for adherence to the treatment plan, adverse effects of therapy (for those on medications), and response to therapy (eg, the achievement of target goals and the occurrence of core symptoms)



- Choice of medication for treatment of ADHD symptoms in TSC or comorbid ASD or anxiety may not be straight forward
- Generally, first line treatment of ADHD is with Stimulants ie: Methylphenidate (Ritalin/ Ritalin LA/ Concerta) or dexamphetamine, Vyvanse
- This may not be the case in comorbid ASD or anxiety
- Non stimulants used for ADHD symptoms are Clonidine, Guanfacine, Atomoxetine



- Stimulants cause decreased appetite, weight loss, insomnia, anxiety, emotional lability. Can enhance tics
- Guanfacine and clonidine causes weight gain and drowsiness. Clonidine causes more drowsiness than guanfacine
- Risperidone and Aripiprazole cause weight gain and increased appetite
- Risperidone increases prolactin levels
- If on Risperidone and aripiprazole then metabolic blood tests are required



Treatment modalities

Behavioural interventions - include modifications in the physical and social environment that are designed to change behaviour using rewards and nonpunitive consequences

- positive reinforcement
- time-out
- response cost (withdrawing rewards or privileges when unwanted or problem behaviour occurs)
- token economy (a combination of positive reinforcement and response cost)



Parent-child behavioural therapy is aimed at improving parent-child relationships through enhanced parenting techniques

Behavioural interventions are most effective if parents understand the principles of behaviour therapy (ie, identification of antecedents and altering the consequences of behaviour) and the techniques are consistently implemented



Behaviour therapy and environmental changes that can be used by parents or teachers to shape the behaviour of children with ADHD include:

- Maintaining a daily schedule
- Keeping distractions to a minimum
- Providing specific and logical places for the child to keep his or her schoolwork, toys, and clothes
- Setting small, reachable goals
- Rewarding positive behaviour (eg, with a "token economy")
- Identifying unintentional reinforcement of negative behaviours
- Using charts and checklists to help the child stay "on task"
- Limiting choices
- Finding activities in which the child can be successful (eg, hobbies, sports)
- Using calm discipline (eg, time out, distraction, removing the child from the situation)

Standardized training programs for parents of preschool children with disruptive behaviour disorders (including symptoms of ADHD) that have been evaluated in systematic reviews include:

- Incredible Years Parenting Program (www.incredibleyears.com/index.asp)
- New Forest Parenting Program (specifically designed to address ADHD symptoms)
- Positive Parenting Program (Triple P)
- Parent-Child Interaction Therapy
- Helping the Noncompliant Child

Other treatment modalities

- Pharmacotherapy
- Combined therapy (behaviour therapy and medication)
- School Based Interventions (Classroom modifications and accommodations may include having assignments written on the board, sitting near the teacher, having extended time to complete tasks, being allowed to take tests in a less distracting environment, or receiving a private signal from the teacher when the child is "off-task.")
- The school also may provide behaviour management programs that target ADHD symptoms and/or enhanced academic and social function



Thank you

