

Neurodevelopmental Disorders in Children with TSC – Surveillance, Screening, Diagnosis and Intervention

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The Plan

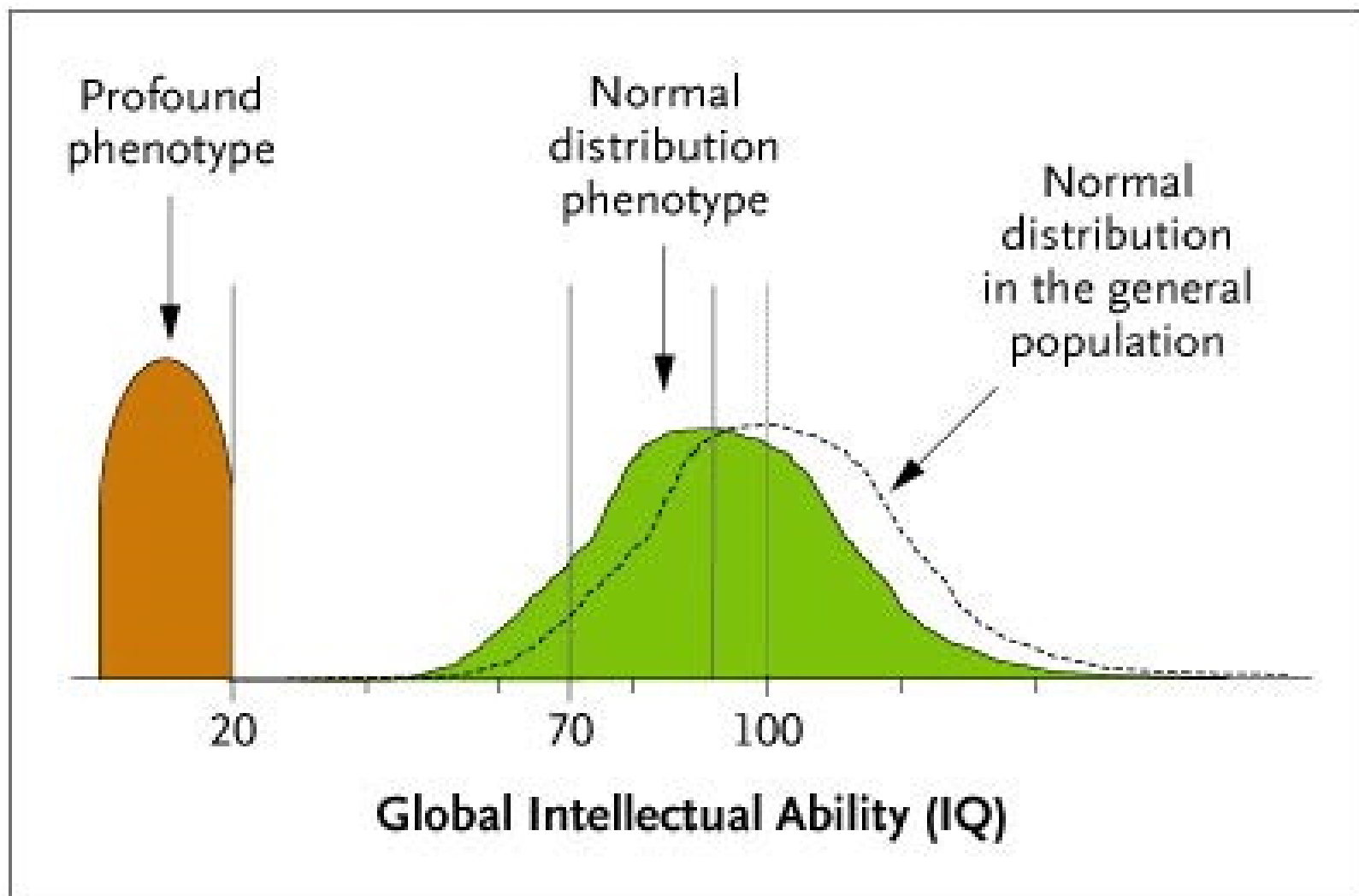
- 15 minutes – Introduction, Neurodevelopmental Disorders in TSC, Typical Development, Developmental Delay
- 5 minutes – Break Out Group 1
- 15 minutes – Screening, Surveillance and Assessment
- 5 minutes – Break Out Group 2
- 10 minutes – Early Intervention
- 10 minutes – Questions and Discussion

TSC and Developmental Disorders

TSC is associated with an array of behavioural and neuropsychiatric manifestations

- Developmental Delay/Intellectual Disability
- Autism Spectrum Disorder
- Attention Difficulties and ADHD
- Memory and Executive Functioning
- Specific Learning Disorders

Great variability in occurrence between individuals



Child Development

- Progressive acquisition of skills over first 4 to 6 years of a child's life
- Stepwise
- Results from the interaction between the **child and their environment**
- Relates to the changes in the child's ability to **move, perform fine movements with their hands, communicate, learn new knowledge, self care and interact with others**



Developmental Domains

Four main domains/areas:

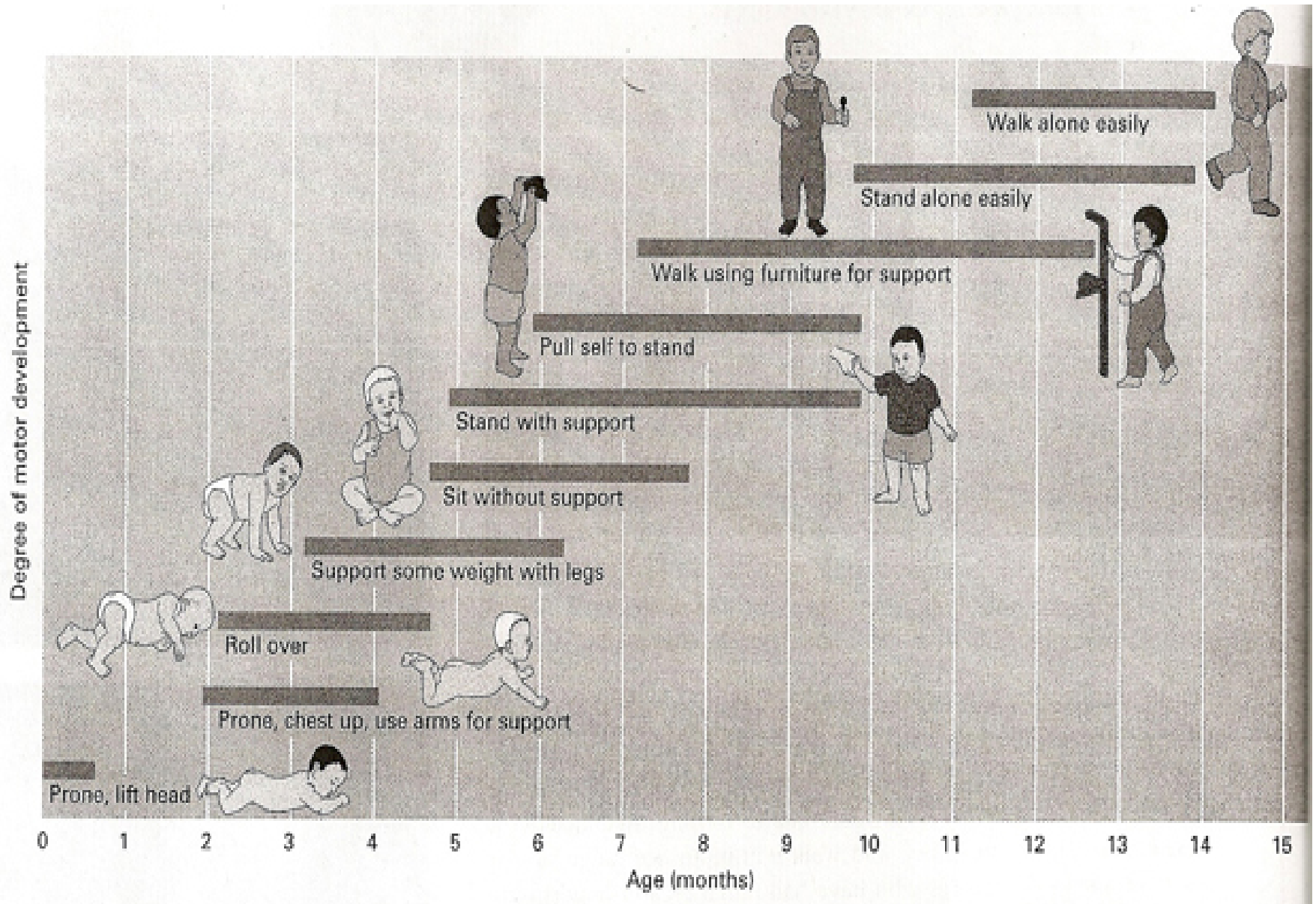
- **Gross motor** - head control, sitting, rolling, crawling, walking, jumping, hopping, skipping, bike riding and ball skills
- **Fine motor** (hand/eye coordination) - attentiveness, handling objects - pencil, scissor, building blocks, threading beads, puzzles and problem solving



Developmental Domains

- **Communication** - responds to sounds, cooing, babbling, single words, comprehension (not compliance) of instructions (1 to 2-3), more complex sentences, understanding concepts
- **Personal social** (self care and socialization skills) - feeding, dressing, grooming, interaction with others, play

Gross Motor Developmental Milestones



Developmental Delay

The first 5 years of a child's life is a period of rapid growth and development where they progress from newborn infants to school ready children with independent mobility, communication and self care skills and are ready to learn.

Developmental delay is the failure to develop those skills in the expected time frame.

Developmental Delay

- Only applies to children under 5 years
- May be confined to one or more developmental domains
- May include all developmental domains – **Global Developmental Delay**

Developmental Flags

- Regression at any age
- Not rolling by 6 months
- Not sitting by 8 months
- Not crawling by 10 months
- No babble by 12 months
- No pointing by 12 months
- No gestures by 12 months
- Not consistently turning to name by 12 months
- No single words by 16 months
- Not walking by 18 months
- No two word combinations by 24 months

Break Out Group 1

- **Where are you getting your developmental and TSC-specific information from?**
- **What Good and Bad sources of information have you found?**

Surveillance, Screening and Assessment

Terminology

- **Surveillance:** Process of recognising children who may be at risk of developmental disorders.
- **Screening:** Use of standardised tools to identify and refine that recognised risk.
- **Evaluation:** A complex process aimed at identifying specific developmental disorder(s).

Screening, Surveillance and Assessment

Assessment

Screening

Universal Surveillance



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Personal Health Record – Surveillance and Screening

Age points:

6 months

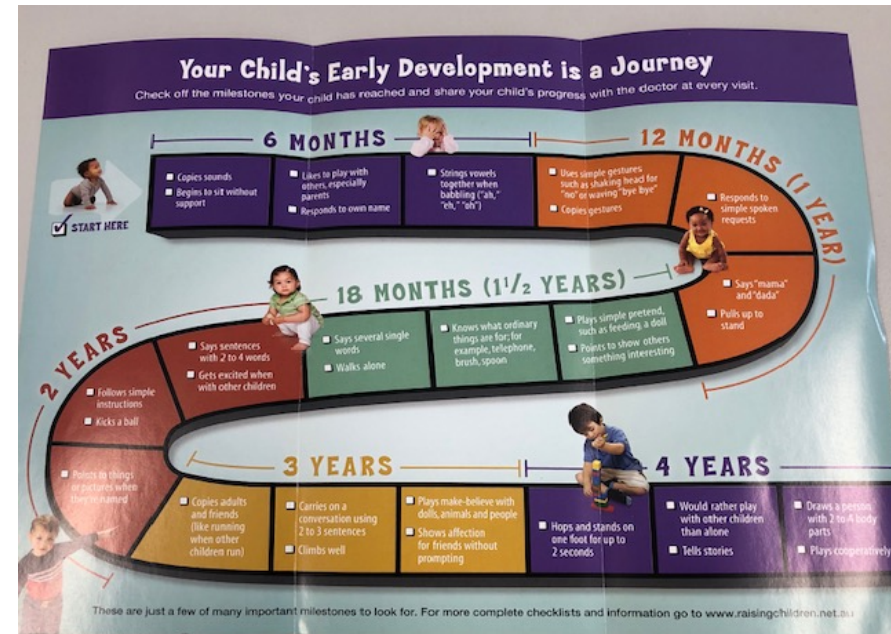
12 months

18 months

2 years

3 years

4 years



<https://www.health.nsw.gov.au/kidsfamilies/MCFhealth/Pages/learn-the-signs.aspx>

Assessments

- Development – global or specific
- Intellectual ability
- Functional ability
- Behaviour
- Learning/academic development

2005 Consensus Guidelines for the assessment of cognitive and behavioral challenges in TSC

- Infancy: Birth - 12 months
- Toddler: 1 year - 2 years 11 months
- Preschool: 3 years to school entry
- Early school years: 6 - 8 years
- Middle school: 9 -12 years
- Adolescence: 13 -16 years
- Adult (follow up): frequency not specified

THE TAND CHECKLIST

Lifetime version (TAND-L)

Tuberous Sclerosis Complex (TSC) is associated with a range of neuropsychiatric disorders which we refer to as **TAND (TSC-Associated-Neuropsychiatric-Disorders)**. All people with TSC are at risk of having some of these difficulties. Some people with TSC have very few, while others will have many of them. Each person with TSC will therefore have their own TAND profile, and this profile may change over time. This checklist was developed to help clinical teams, individuals with TSC and their families

a) screen for TAND at every clinic visit and b) prioritize what to do next.

Instructions for use

The TAND Checklist was designed to be completed by a clinician with relevant knowledge and experience in TSC, in partnership with individuals with TSC or their parents/carers.

The Checklist should take about 10 minutes to complete.

Where individuals answer YES to an item, the clinician should explore the difficulty in sufficient detail to help guide decisions about further evaluation or treatment. All items should be completed.

About the interview

Name of TSC Subject: DOB: dd/mm/yy Age:

Name of Interviewer: Date of interview: dd/mm/yy

Name of interviewee: Self / Parent / Carer / Other (circle)

Let's begin

As you will know, the majority of people with TSC have some difficulty in learning, behaviour, mental health, specific aspects of their development and so on. We are going to use this checklist to help us check for these kinds of difficulties. I am going to ask you a number of questions.

Some may be directly relevant; some might not be relevant at all. Just answer as best as you can.

At the end I will check to see if there are any additional difficulties we didn't talk about.

For parents/carers of individuals with TSC, please start with question 1.

For individuals with TSC who complete this about themselves, please start with question 3.

01

Let's begin by talking about [subject]'s development to get a sense of where they are at. How old was [subject] when he/she:

- | | | |
|---|---------------------------|-----------------------------------|
| a. First smiled? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| b. Sat without support? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| c. Walked without holding on? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| d. Used single words other than "mama" or "dada"? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| e. Used two words/short phrases? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| f. Was toilet trained during the day? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |
| g. Was toilet trained at night? | Age: <input type="text"/> | Not yet: <input type="checkbox"/> |



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Consensus clinical guidelines for the assessment of TSC-Associated Neuropsychiatric Disorders (TAND) All individuals with TSC should be screened for TAND at least annually and more detailed evaluations should follow from screening. In addition, all individuals with TSC should have a comprehensive formal evaluation at key developmental timepoints as outlined below in this table (de Vries et al., 2005; Krueger et al., 2013).

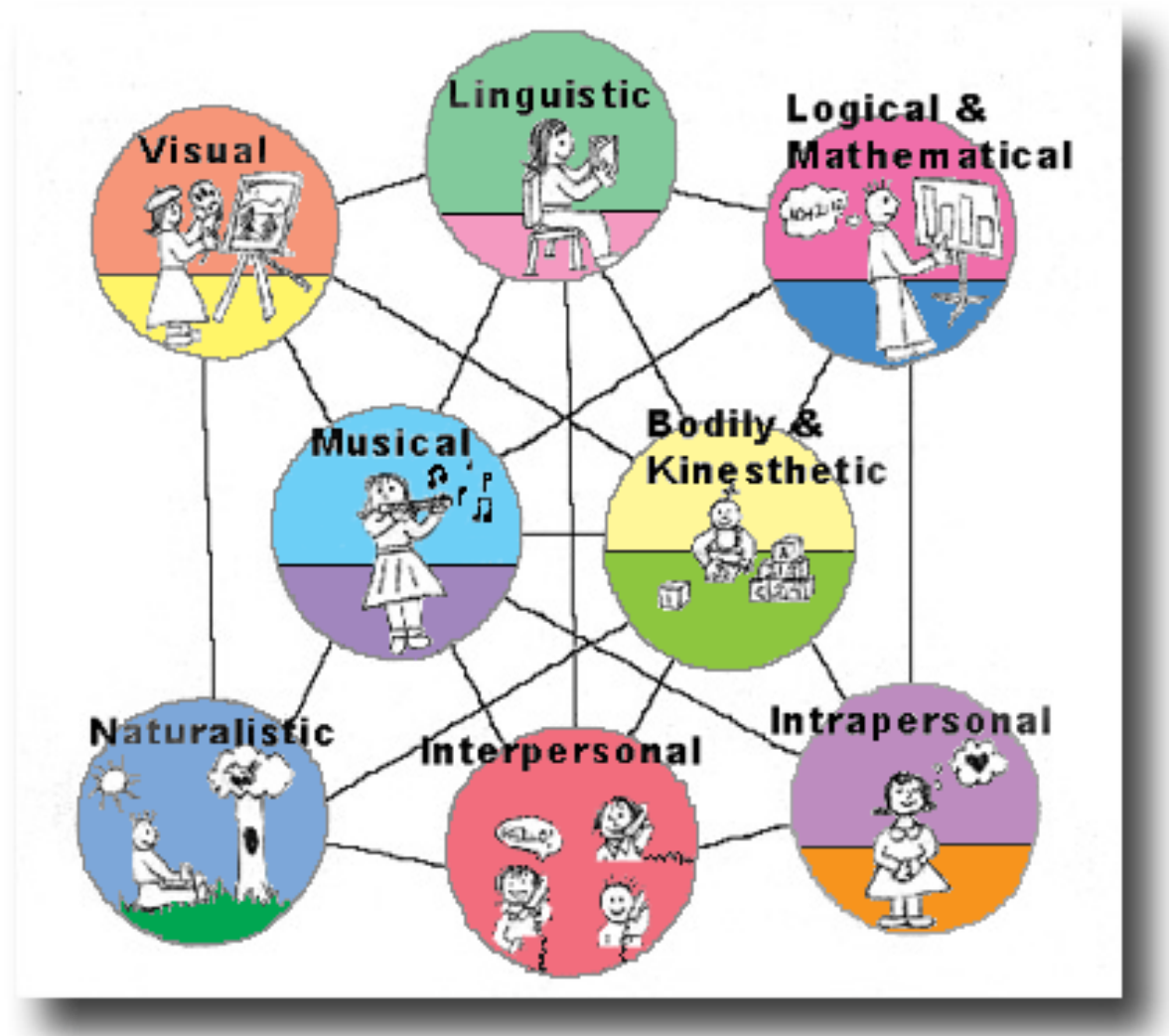
| STAGE | AGE RANGE | GENERAL PURPOSE OF ASSESSMENT | GENERALAREASTO ASSESS | AREAS OF PARTICULAR CONCERN IN TSC | BEHAVIORAL, PSYCHIATRIC, AND ACADEMIC DISORDERS OF PARTICULAR CONCERN IN TSC |
|--------------------|---------------------|---|---|--|--|
| At diagnosis | | Initial assessment of cognitive and behavioral profile | As listed for chronological age | | |
| Infancy | Birth – 12 mos | To perform a baseline assessment for regular monitoring of development | Global standardized assessment of infant development | Impact of seizure onset and treatment on development | |
| Toddler | 1y – 2y11m | To identify early developmental disorders Specific skills: • Gross and fine motor skills • Social-communication skills | Global intellectual ability and adaptive behaviors | Quality of eye contact, joint attention, reciprocity | Autism and autism spectrum disorders (ASD) Severe aggressive outbursts Severe sleep problems |
| Pre-school | 3 y to school entry | Evaluation of cognitive and behavioral profile to ensure the provision of appropriate educational programs | Global intellectual ability Specific neuropsychological skills: • Receptive and expressive language • Social-communication skills • Attentional and executive skills • Visuospatial skills • Motor skills | Uneven profile of abilities Poor expressive language Poor reciprocity, peer interaction Poor regulation of affect and impulse Poor bilateral coordination | Autism and ASD ADHD and related disorders Self-injurious behavior |
| Early school years | 6y-8y | Monitoring the child's ability to make appropriate educational progress | Global intellectual abilities Specific neuropsychological skills: • Receptive and expressive language • Social-communication skills • Memory • Attentional and executive skills • Visuospatial skills • Motor skills | Best time to establish baseline to assess whether specific cognitive skills and academic performance are discrepant from global intellectual abilities Poor expressive language and word retrieval Rote learning difficulties Selective attention, sustained attention difficulties | Academic difficulties (reading, writing, spelling, mathematics) ADHD and related disorders Peer problems Aggressive behaviors |

The consensus clinical guidelines for the assessment of TAND (continued)

| STAGE | AGE RANGE | GENERAL PURPOSE OF ASSESSMENT | GENERAL AREAS TO ASSESS | AREAS OF PARTICULAR CONCERN IN TSC | BEHAVIORAL, PSYCHIATRIC, AND ACADEMIC DISORDERS OF PARTICULAR CONCERN IN TSC |
|---------------------|-----------|---|---|---|---|
| Middle school years | 9y-12y | Comprehensive review of child's abilities, specific learning difficulties, and behavioral problems in preparation for transition to secondary education | Global intellectual abilities Specific neuropsychological skills: • Receptive and expressive language • Social-communication skills • Memory skills • Attentional and executive skills | Subtle deficits of social communication, unusual interests Poor working memory, episodic memory Planning, organizational abilities, multi-tasking difficulties | High functioning ASD/Asperger's Peer problems Academic difficulties (reading, writing, spelling, mathematics) |
| Adolescence | 13y-16y | Determining individual needs and the support required for transition into adulthood | Global intellectual abilities Specific neuropsychological skills: • Attentional and executive skills • Vocational assessment with knowledge of cognitive strengths and weaknesses • Adaptive behavior and daily living skills | Poor judgement, decision-making | Depressive disorders Anxiety disorders Peer problems Epilepsy-related psychotic disorders |
| Adults | 18y+ | Newly diagnosed adults: Assessment of cognitive, behavioral and vocational profile, determining bio-psycho-social needs | Global intellectual abilities Specific neuropsychological skills: • Attentional and executive skills • Memory skills | Difficulty with integrational skills Working memory, episodic memory problems | Depressive disorders Anxiety disorders Epilepsy-related psychotic disorders |
| Adults (follow-up) | 18y+ | Monitoring for emergence of psychiatric problems or changes in existing cognitive and behavioral profile | Dependent adults: • Annual review of social care needs and support Independent adults: • Vocational advice • Genetic counseling as appropriate • Review if problems arise | Pay particular attention to change in cognitive abilities or behavior Pay particular attention to change in cognitive abilities, vocational performance and behavior | Depressive disorders Anxiety disorders Epilepsy-related psychotic disorders |

The table shows the time points recommended for evaluation and the goals of evaluation and lists specific areas of concern for each age group. Table reproduced with permission from de Vries et al. (2005) updated by de Vries 2014. Note: Many features listed in these columns can present at any age, but are listed here at stages most commonly associated with the emergence of such difficulties in TSC.

Howard Gardner's Types of Intelligence



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Early Identification of Developmental Delay/Disorders

Early identification is critical to the well being of children and their families

- Intervention
- Identification and management of medical co-morbidities
- Family support
- Genetic counselling

Developmental Vulnerability

Children at risk of developmental delay by virtue of their:

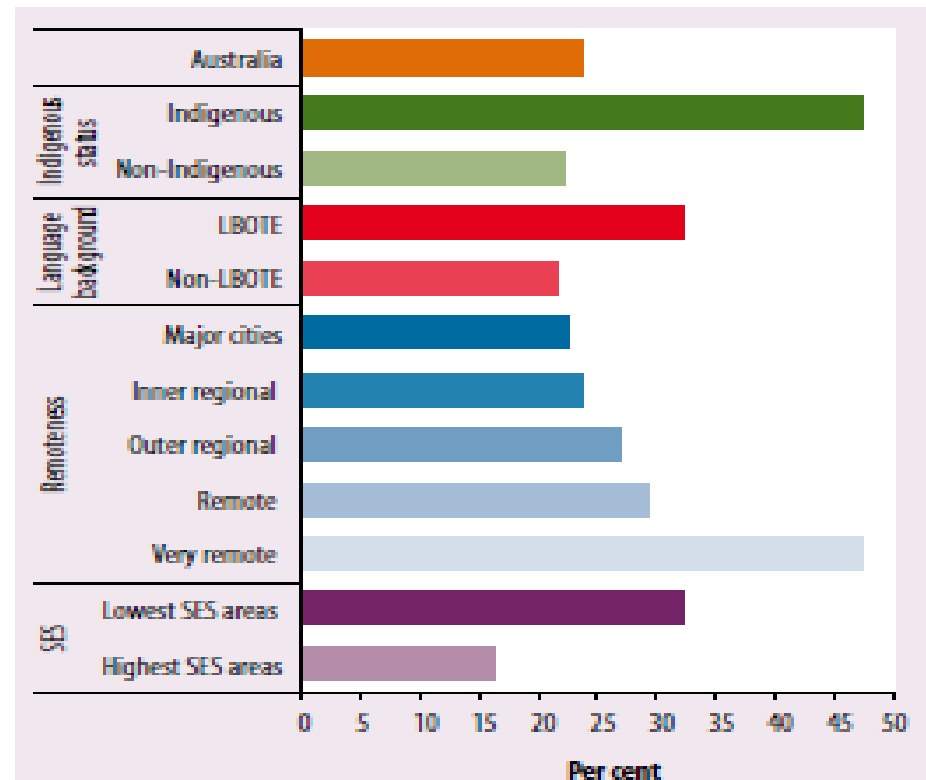
- Genetic factors
- Medical factors
- Psychosocial factors

Developmental Vulnerability in Australian Children

Depending on the tool and the population between 10- 25% of children in high income countries are developmentally vulnerable.

They do not have the skills required to flourish at school.

How does developmental vulnerability vary across population groups?



Notes

1. LBOTE refers to language background other than English.
2. Refer to Appendix 2: Methods for explanation of remoteness areas and socioeconomic status (SES).

Source: Australian Early Development Index unpublished data; Table A1.14b.

Figure 14.2: Children developmentally vulnerable on one or more domains of the AEDI, by selected population groups, 2009

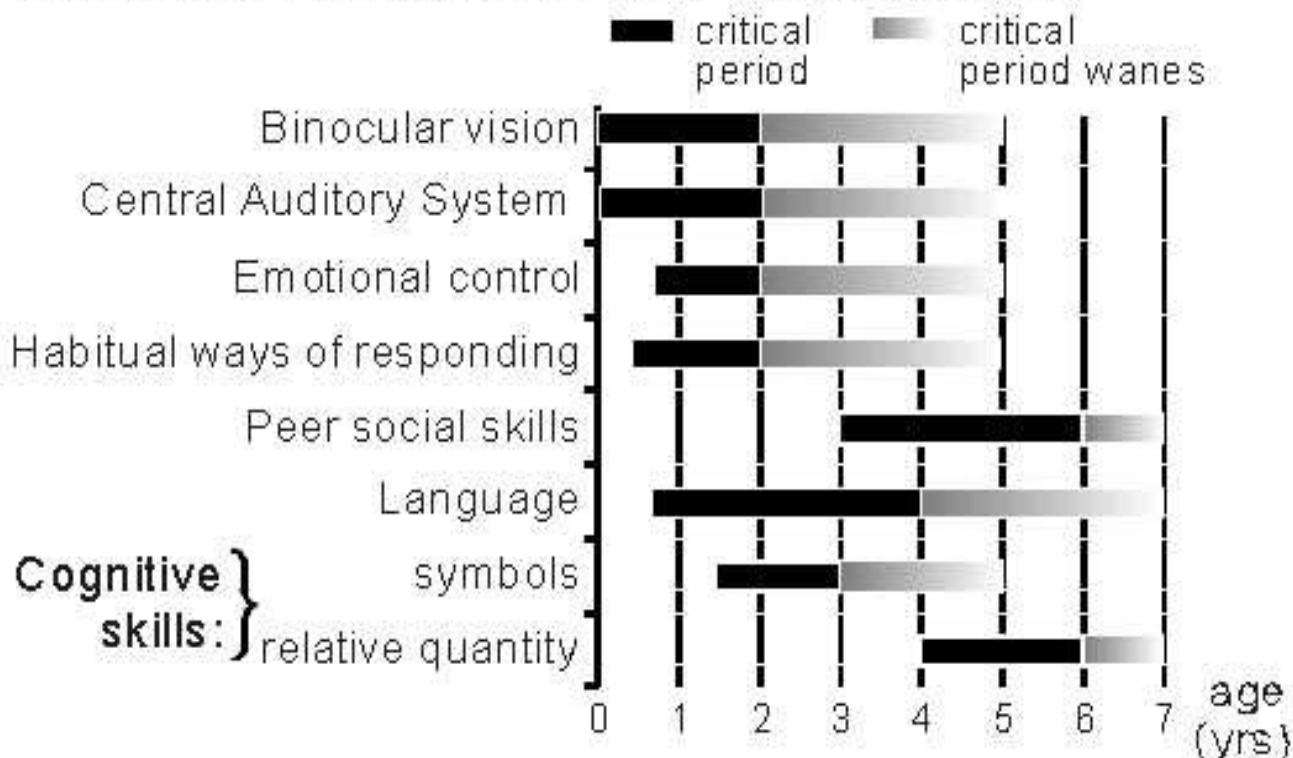
Early Intervention

- Plasticity of early brain development
- Critical periods of development
- Prevention of secondary disability
- Enhanced functioning of the child and family
- Provision of family support and resources

Sensitive or critical periods of development

- Periods in development when a number of factors, brain development, sensory input and environmental factors, act together to promote or enhance the acquisition of a particular skill.
- If some factor prevents this skill development at the appropriate time, subsequent learning may be more difficult and imperfect.
- Examples:
 - Development of visual pathways; effect of lack of use of vision in one or both eyes.
 - Language development: acquisition of primary language; effect of undiagnosed early hearing impairment.

Sensitive Periods for Early Development



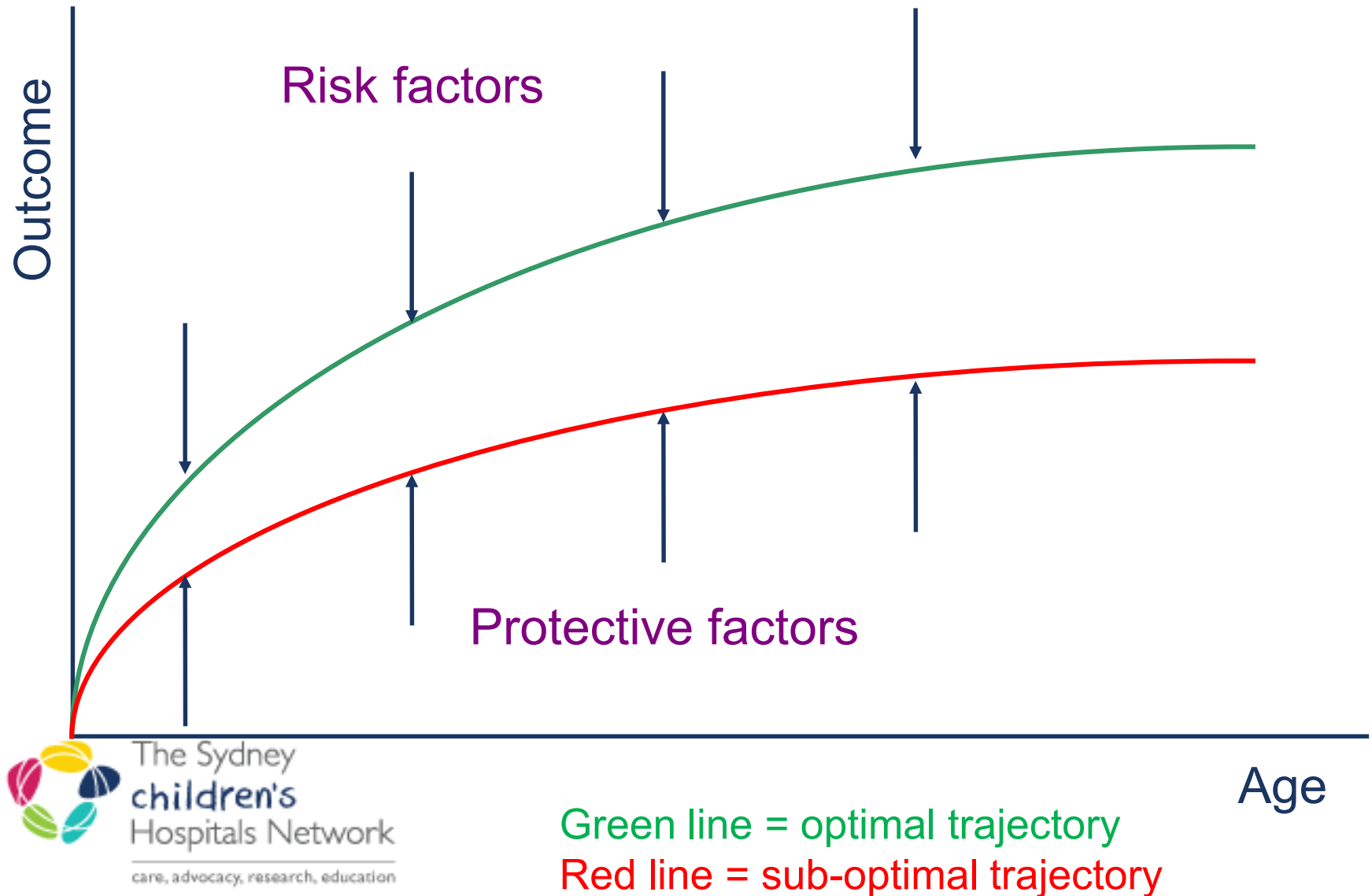
Source: adapted from Begley, 1996, pp. 68-69
 "Zero to Six: The Basis for School Readiness"
 R-97-8E, Billie Doherty May 1997
 Applied Research Branch Strategic Policy, Human
 Resources Development Canada



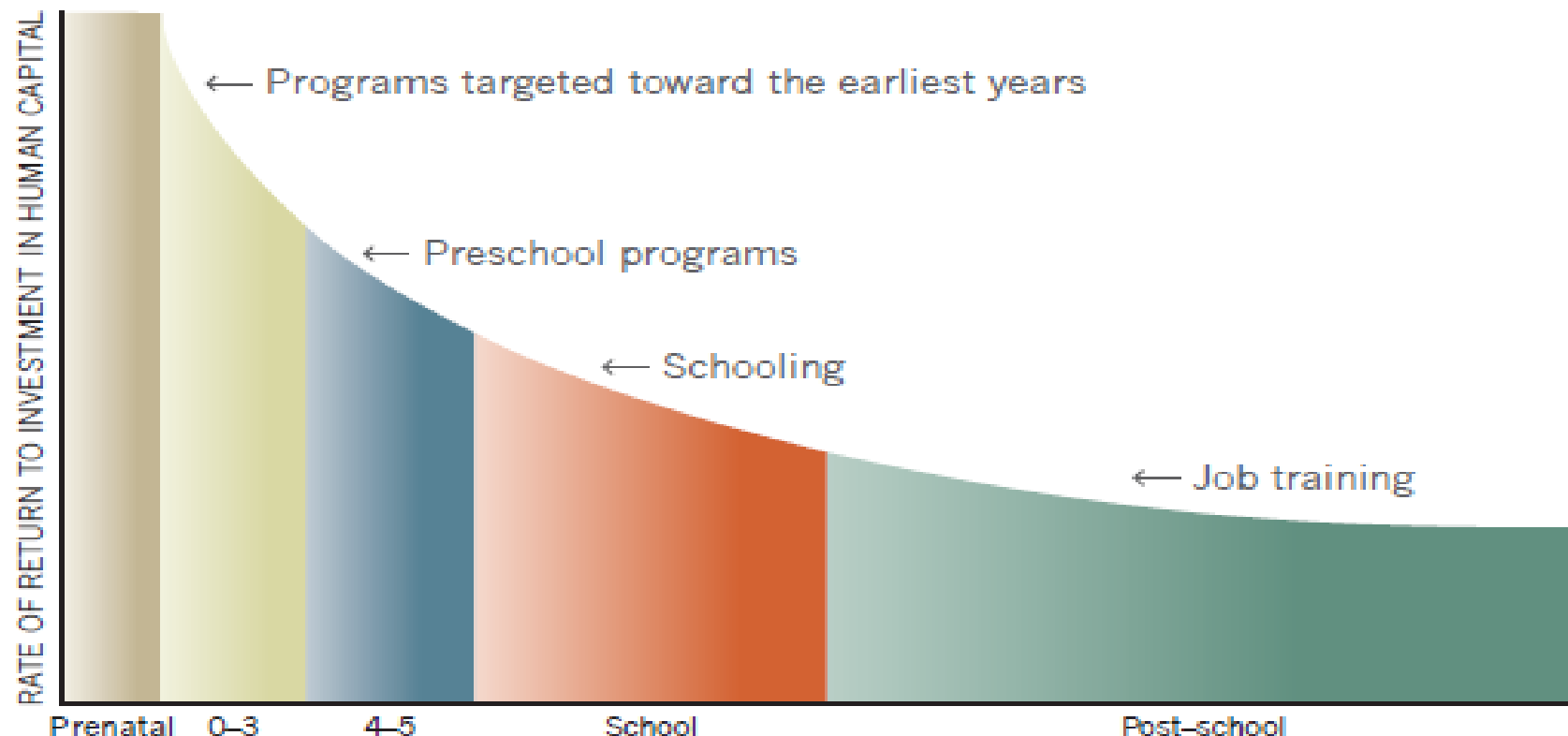
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Optimising Outcomes



Returns to a Unit Dollar Invested



Heckman, James J. (2008). "Schools, Skills and Synapses," *Economic Inquiry*, 46(3): 289-324



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[https://heckmanequation.org/assets/2013/07/F_HeckmanDeficitPieceCUSTOM-
Generic_052714-3-1.pdf](https://heckmanequation.org/assets/2013/07/F_HeckmanDeficitPieceCUSTOM-Generic_052714-3-1.pdf)

Break Out Group 2

- **Are you using the universal surveillance system?**
- **Have you had any assessments?**
- **How was the experience?**
- **Was it easy to get an assessment?**

Intervention



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Early Interventions

Universal

Targeted



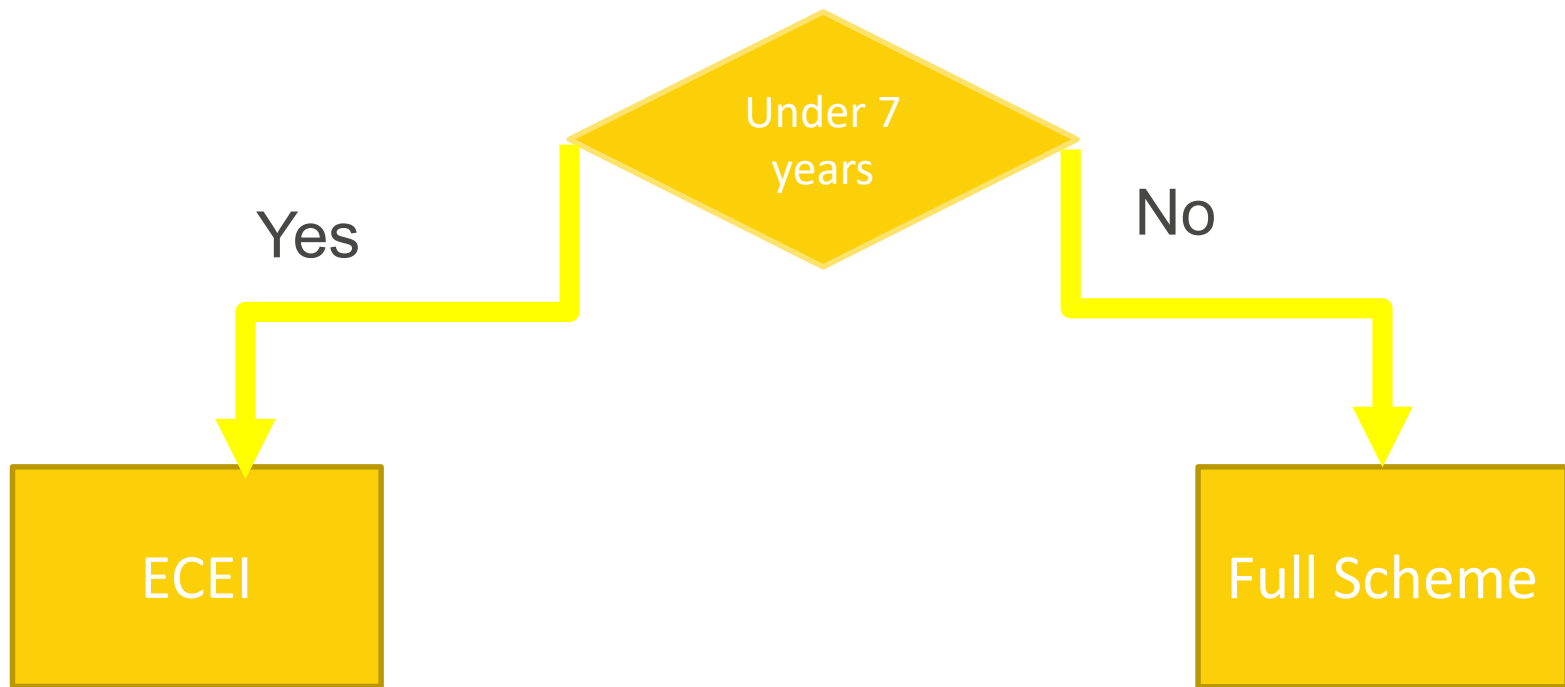
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Early Childhood Therapy

- Speech Pathologist
- Occupational Therapist
- Physiotherapists
- Early Childhood Educators
- Key Workers
- Psychologists

NDIS - referrals



ECEI Partners – NSW regions

Lifestart - Central Coast, Illawarra Shoalhaven, South Eastern Sydney, Sydney and Nepean Blue Mountains

- <http://www.lifestart.org.au/> ph: 1800 953 390

Northcott – Hunter New England, Mid North Coast, Northern NSW & Western Sydney Service area

- <https://northcott.com.au/ECEI> ph: 1800 818 286

Mission Australia – Far West and Western NSW

- <https://www.missionaustralia.com.au/what-we-do/disability-inclusion-and-support>
ph: (02) 6884 5526

EACH – South Western Sydney, Southern NSW Service area

- <http://www.each.com.au/service/early-childhood-early-intervention/> ph: 1300 003 224

Intereach – Murrumbidgee, Goulburn, Lodon, Mallee Service areas

- <http://www.intereach.com.au/support/disability/> ph: 1300 488 226

Cerebral Palsy Alliance - Northern Sydney

- <https://www.cerebralpalsy.org.au/services/ndis-early-childhood-early-intervention-partners-in-the-community/> ph: 1300 888 378

Early Education



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NSW DEPARTMENT OF EDUCATION FUNDED PROGRAMS

Font



Educational supports for children:

- * with developmental delay/ disability
- * 2 – 4 years of age not accessing preschool or child care centre



Start Strong Pathways – Sydney
Metropolitan area - Lifestart
ph: 1800 111 753



Building Tots Early Childhood
Development Program – Sydney
and Rural Areas - Aspect
ph: 02 8977 8341



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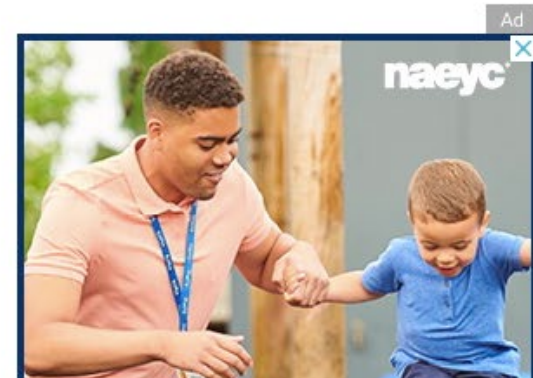
What Does a High-Quality Preschool Program Look Like?



What Does a High-Quality Preschool Program Look Like?

Teachers focus on these areas to help children learn best.

Creating a Caring Community of Learners



Parenting



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Milestones



Share a
Summary



Get Tips &
Activities

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🏠 Child Development

Child Development +
BasicsDevelopmental +
Screening**Positive Parenting —
Tips**

Infants (0-1 year)

Toddlers (1-2 years)

Toddlers (2-3 years)

Preschoolers (3-5
years)Middle Childhood
(6-8 years)Middle Childhood
(9-11 years)

Positive Parenting Tips

[Español \(Spanish\)](#)

As a parent you give your children a good start in life—you nurture, protect and guide them. Parenting is a process that prepares your child for independence. As your child grows and develops, there are many things you can do to help your child. These links will help you learn more about your child's development, positive parenting, safety, and health at each stage of your child's life.



APP & RESOURCES

LOVE Talk SING Read PLAY APP

This app has been developed as a Families NSW project in partnership between South Western Sydney Local Health District, South Eastern Sydney Local Health District, Sydney Local Health District, Families NSW St George Child and Family Interagency and Resourcing Parents.

The Love Talk Sing Read Play app contains information for every family to help their child/ren learn and develop.

- Get help on how to help your child/ren learn and develop.
- Add photos of your child/ren and create a memory book.
- Choose your own captions or use the ones provided.
- Be sent reminders on immunisations and blue book checks.
- Use our generic parenting contacts or add your own personalised contacts.
- Download the App, set up your profile and select your language; English, Arabic, Bengali, Chinese or Nepali.



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raisingchildren.net.au

the australian parenting website



Search the site



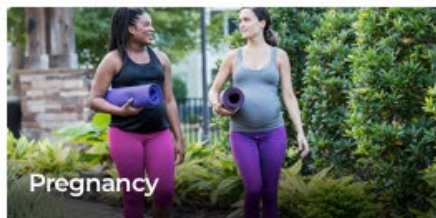
pregnancy & grown-ups ▾

babies & children ▾

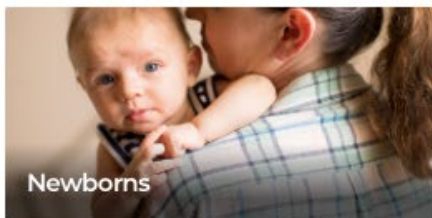
teens ▾

autism & disability ▾

[Read more >](#)



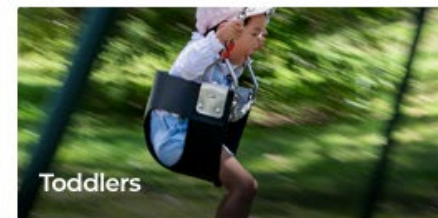
Pregnancy



Newborns



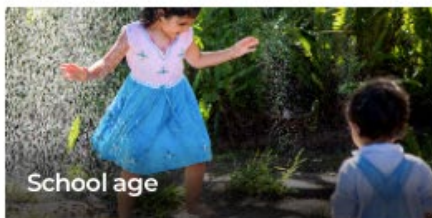
Babies



Toddlers



Preschoolers



School age



Pre-teens



Teens



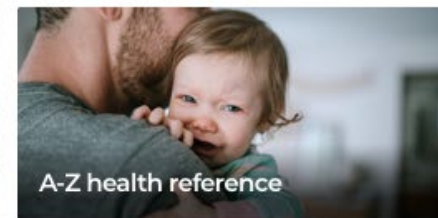
Grown-ups



Autism



Disability



A-Z health reference



Helping children with disability learn through everyday interests and play



Play and children with autism spectrum disorder



Friends and peers: children and teenagers with autism spectrum disorder



Parties and social events: children and teenagers with autism spectrum disorder



Preschoolers making friends



Friends and friendships: 10 frequently asked questions



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Resources for professionals

Professionals, this collection brings together articles, videos and guides related to mental health from across raisingchildren.net.au. Better support the families you work with by quickly and easily accessing our up-to-date and evidence-based content on child, teenage and parent mental health and wellbeing. Get information, download resources or refer parents to raisingchildren.net.au.

Search mental health topics by using the A-Z index or browsing topics below.

A B **C** D E F G H I J K L M N O P Q R S T U V W >



- Calling out and getting out of bed
- Child health - services
- Child protection, health and safety services
- Child safety - services
- Child sexual abuse: help and support for children
- Child sexual abuse: helplines and services
- Child sexual abuse: talking to children 0-11 years

- Childhood sexual behaviour - when to be concerned
- Children's impact on your relationship
- Choosing mental health services for teenagers
- Communication - tips for parents on talking to each other
- Community activities: getting teenagers involved
- Confidence in teenagers



Any Questions



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