

# SURVEILLANCE AND SCREENING IN AUTISM



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# AUTISM SPECTRUM DISORDER (ASD)

- A neurodevelopmental disorder that has diverse etiologies
- Characterised by impairments in social communication and social interaction, as well as the presence of repetitive and restricted behaviours, interests and activities
- Characterised by heterogeneity & chronogeneity



- **Prevalence of ASD:**
  - Significant increase reported since start of century
  - Affects 1 in 54 children in USA ( $\pm$  2%) – CDC  
(CDC estimates are calculated only for children aged 8 yrs)
  - Global – between 1 and 2 %
  - Absence of data in sub-Saharan Africa & South Africa
- **Gender:**
  - Male-to-Female ratio 4.3:1
  - Decreases in ratio have emerged
- **Average Age of Diagnosis:**
  - Sx can be identified by 12 months
  - Reliable Dx by 24 months
  - Median age of diagnosis 4 yrs 3 months (51 months)

The background of the image is a dense grid of numerous small, circular, colorful patterns. These patterns are highly diverse, featuring various colors like red, blue, green, yellow, and purple, and different shapes and textures, including spirals, dots, and abstract forms. This visual metaphor represents the vast and unique range of experiences and traits within the autism spectrum.

**Autism Spectrum:  
everyone is different**

# ASD DEFIES GENERALISATION

## Measured Intelligence:

Severely Impaired \_\_\_\_\_ Gifted

## Social Interaction:

Aloof \_\_\_\_\_ Passive \_\_\_\_\_ Active but odd

## Communication:

Nonverbal \_\_\_\_\_ Verbal

## Behaviors:

Intense \_\_\_\_\_ Mild

## Sensory:

Hyposensitive \_\_\_\_\_ Hypersensitive

## Motor:

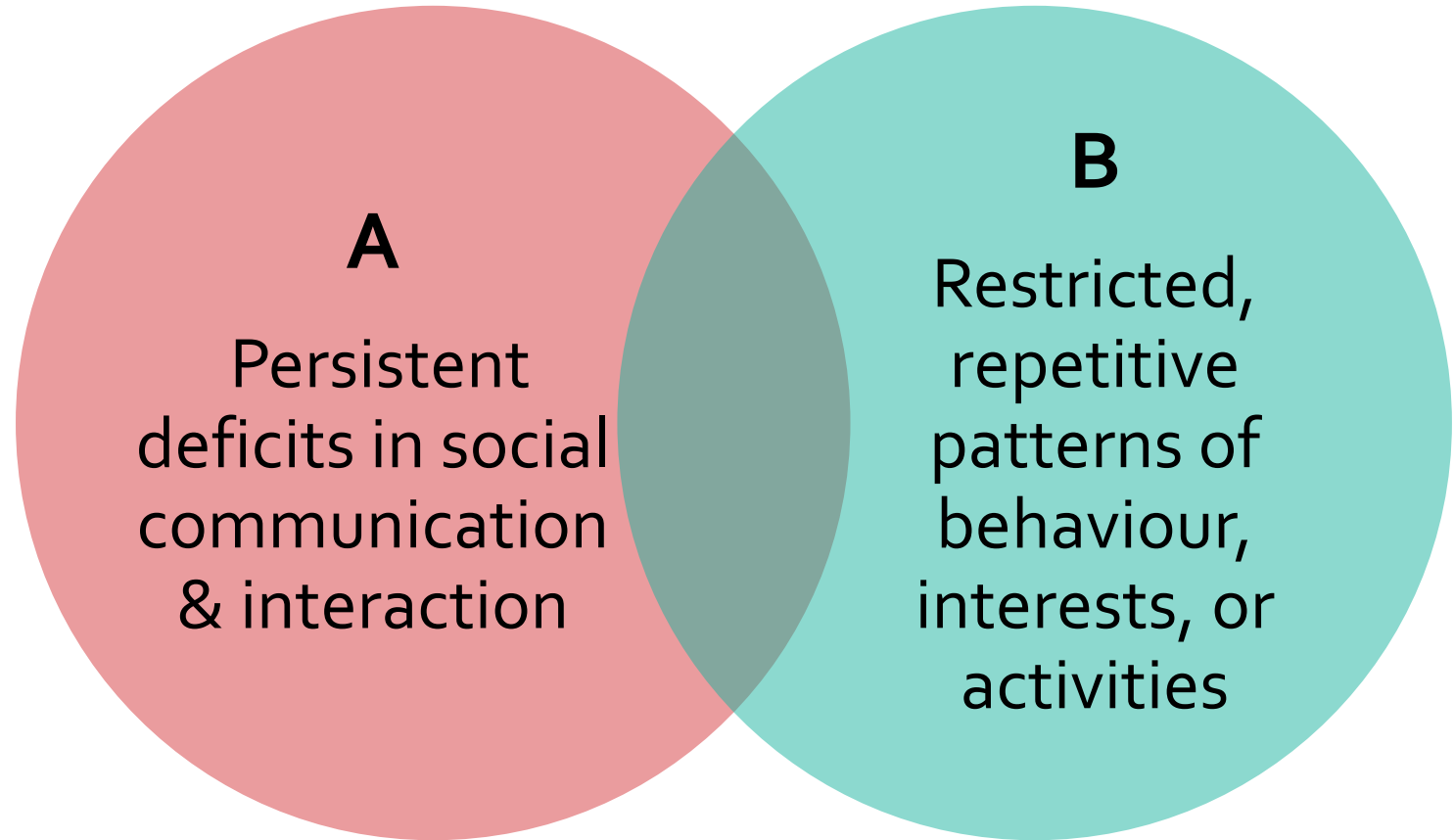
Uncoordinated \_\_\_\_\_ Coordinated



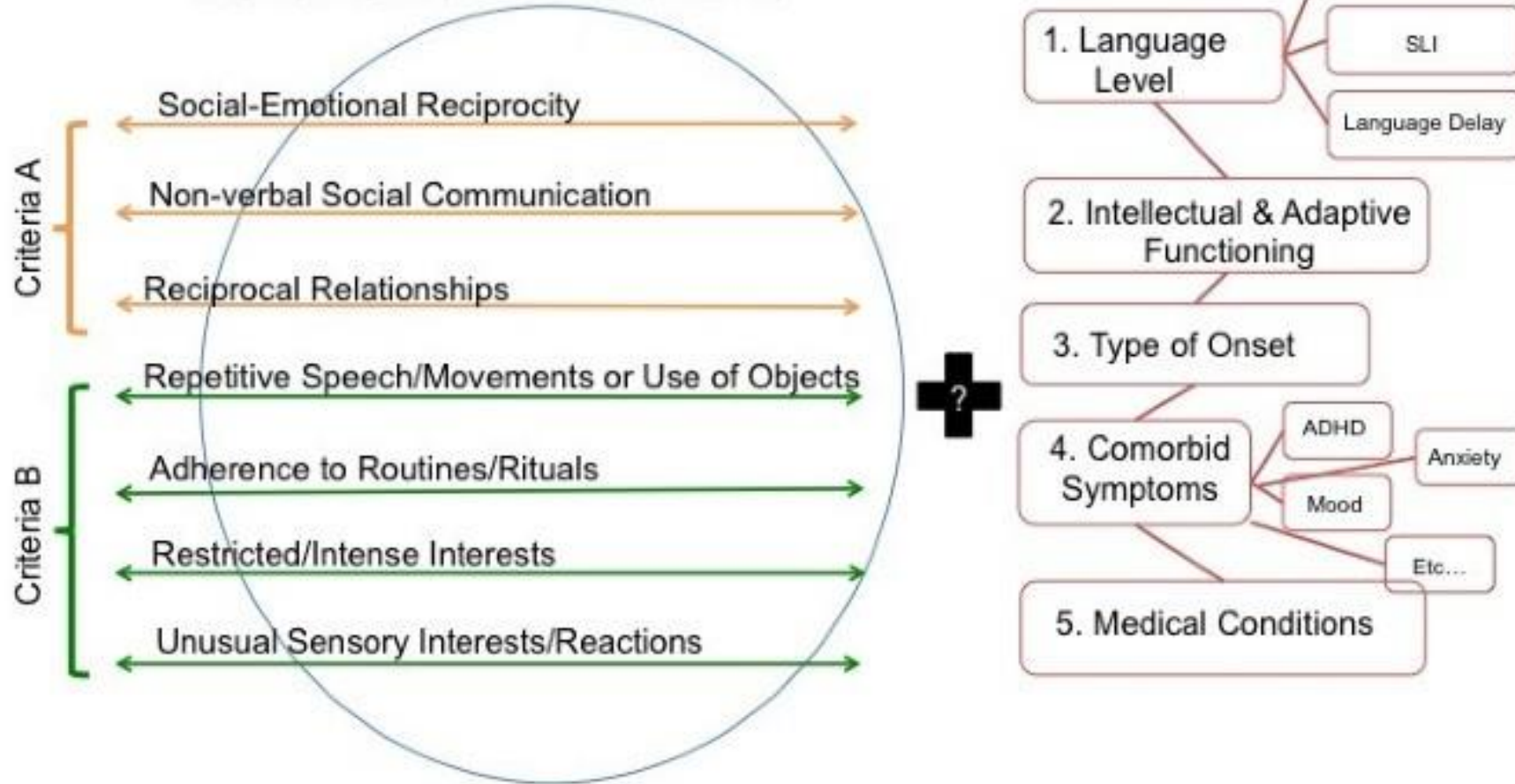
- DSM-5 → ultimate diagnostic reference, but not formal test
- Gold standard for dx of ASD – 2 tests:
  - Autism Diagnostic Interview-Revised (ADI-R)
  - Autism Diagnostic Observation Schedule 2 (ADOS 2)



# DSM-5 DIAGNOSTIC CRITERIA



# AUTISM SPECTRUM DISORDERS





BEFORE 2013, FIVE DIFFERENT SUBTYPES OF AUTISM WERE CLASSIFIED UNDER:

## 1994 - 2013: PERSVASIVE DEVELOPMENT DISORDERS (PDD)

### AUTISM

AUTISTIC DISORDER  
"CLASSIC" AUTISM

### ASPERGER'S

ASPERGER SYNDROME

### CDD

CHILDHOOD DISINTEGRATIVE  
DISORDER

### PDD-NOS

PERSVASIVE DEVELOPMENTAL  
DISORDER - NOT OTHERWISE  
SPECIFIED

### RETT'S

RETT SYNDROME



FOUR OF THESE SUBTYPES HAVE NOW BEEN REPLACED WITH ONE CENTRAL DIAGNOSIS:

## 2013 - PRESENT: AUTISM SPECTRUM DISORDER (ASD)

RATHER THAN A DISTINCTION BETWEEN DIFFERENT SUBTYPES, ASD IS NOW  
DIFFERENTIATED BASED ON THREE LEVELS OF SEVERITY AND SUPPORT NEEDED:

### LEVEL 1

REQUIRING SUPPORT

Problems with inflexibility, poor organization, planning, switching between activities, which impair independence. Poor social skills, difficulty in initiating interactions, attempts to make friends are odd and unsuccessful.



### LEVEL 2

REQUIRING SUBSTANTIAL  
SUPPORT

Marked difficulties in **verbal** and **nonverbal** social communication skills. Markedly odd, restricted **repetitive behaviors**, noticeable difficulties changing activities or focus.



### LEVEL 3

REQUIRING VERY  
SUBSTANTIAL SUPPORT

Severe difficulties in **verbal** and **nonverbal** communication. Very limited speech, odd, **repetitive behavior**; many express their **basic needs** only.

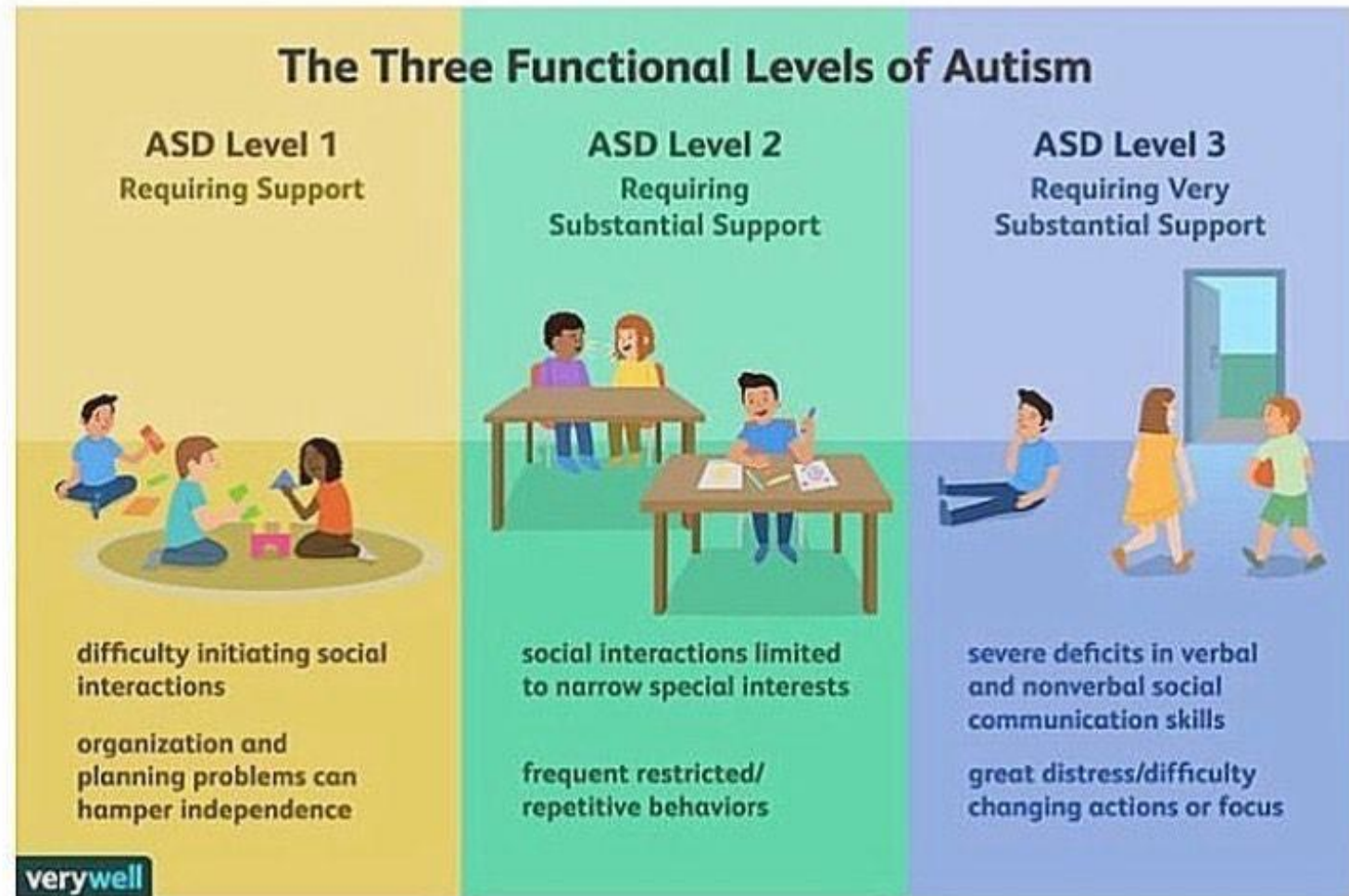


#LIGHTITUPBLUE

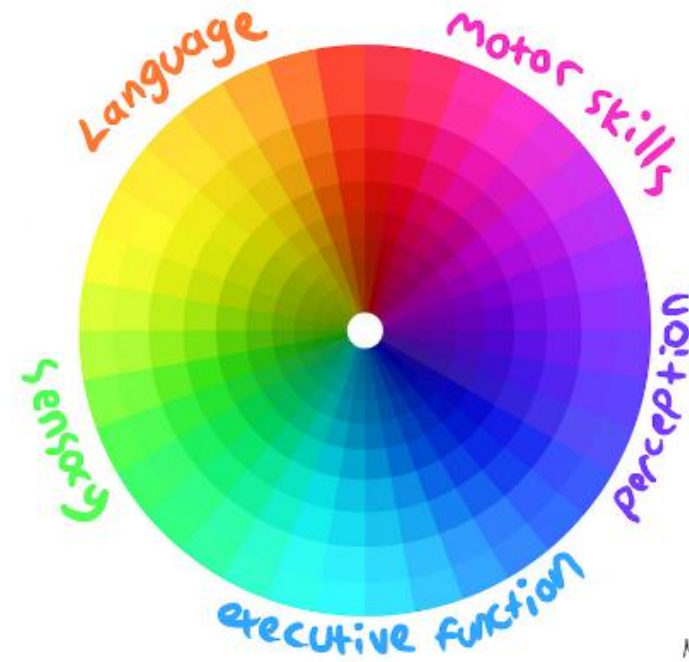
Source: Autism-Europe

infographic.ly

# Levels of Support



What does the autism spectrum look like?



Circular spectrum by  
Rebecca Burgess  
Montage @sciencebase

# DELAYED DIAGNOSIS

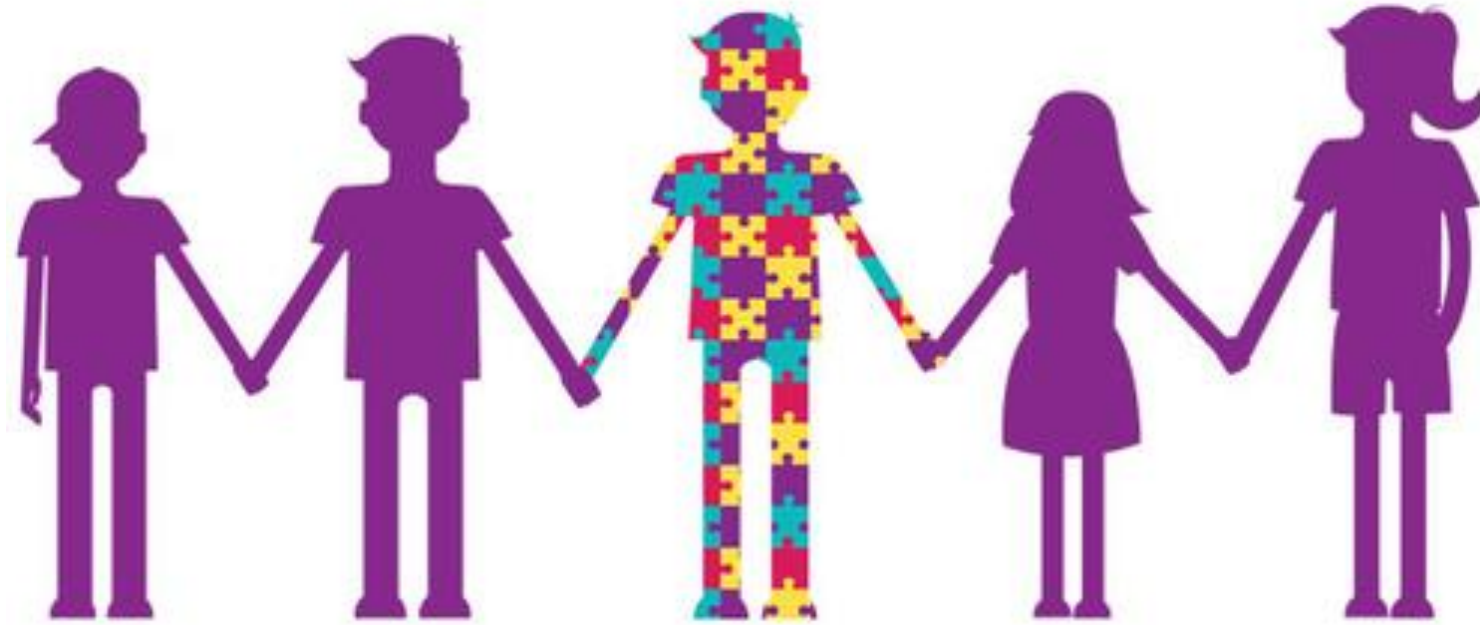


- Mean age of dx has not improved significantly over the past 2 decades despite efforts to educate the public and health professionals
- Delays:
  - Between onset of parental concerns and comprehensive evaluation
  - Between comprehensive evaluation and dx of ASD
- Factors that may delay diagnosis:
  - Less severe Sx
  - Female
  - Co-existing problems
  - Lack of continuity of care
  - Hearing impairment
  - Oversensitivity to pain
  - Language barriers
  - Lower socioeconomic status
  - Living in rural area
  - Attribution of regression of skills to “stressors”(e.g. birth of sibling)
  - Lack of knowledge of child development



Early identification and intervention important

- During 1<sup>st</sup> years of life critical period of synaptogenesis occurs
- Environmental factors & experiences influence both construction of specific neural circuits & elimination of excess synapses
- Early environmental enrichment during 1<sup>st</sup> years of life has been found to alter the trajectory of both biological & behavioural development
- Early intervention effective in improving adaptive behaviour, language & IQ in general and also specific in ASD



# SURVEILLANCE AND SCREENING

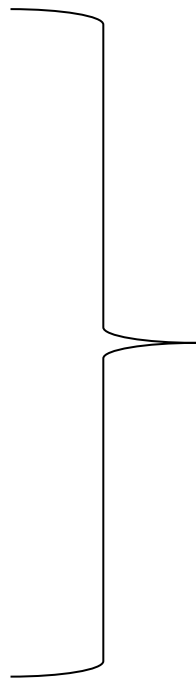
**Developmental surveillance**  
= observation & information gathering



**Developmental screening**  
= brief standardised test  
Determine risk or presence of a problem



**Developmental evaluation**  
= comprehensive evaluation  
Determine diagnosis & plan intervention



**Early developmental intervention**  
= therapeutic intervention to  
optimise developmental outcome



## SURVEILLANCE

Process through which potential risk factors for developmental and behavioural disorders can be identified

Defined as flexible, continuous process in which knowledgeable professionals collect information from multiple sources to identify and address developmental concerns

# What information in the Road to Health Booklet is relevant to Developmental surveillance?

- Child's special care needs (disability)
- Information on feeding and nutrition
- Birth & neonatal information
- HIV status & management
- Growth trends
- Hospital admissions and reasons therefore
- Long term health conditions
- Clinical notes from healthcare contacts
- Health promotion messages
- Developmental screening table

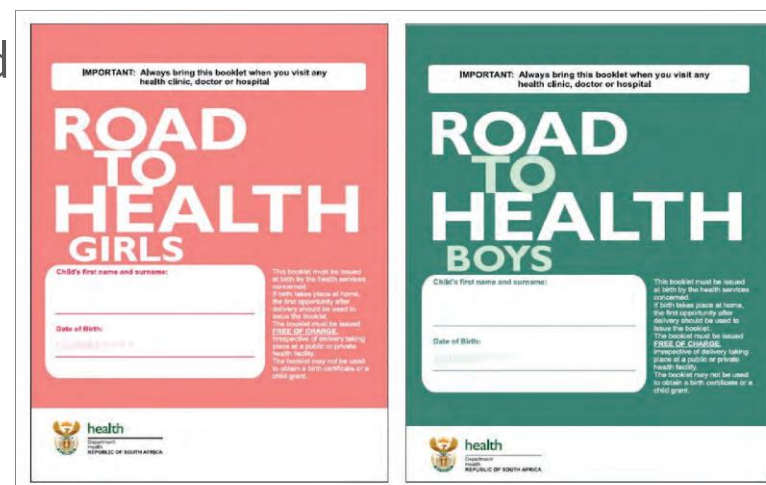
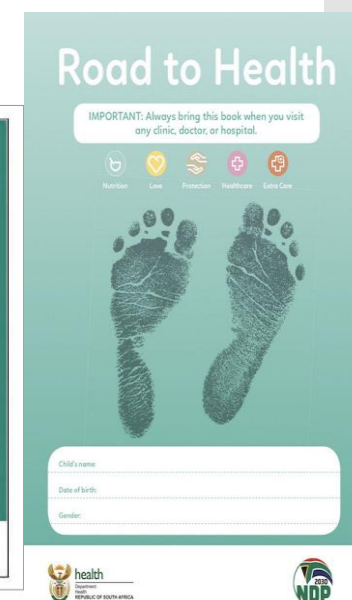


FIGURE 7.A1: Road to Health Booklet





## Developmental screening



8 w. ....	Hearing/ communication	Vision and adaptive	Cognitive/ behaviour	Motor skills	Caregiver concerns
<b>10 weeks</b>					
<b>14 weeks</b>	<input type="checkbox"/> Startles to loud sounds	<input type="checkbox"/> Follows face or close objects with eyes	<input type="checkbox"/> Smiles at people	<input type="checkbox"/> Holds head upright when held against shoulder	
Date ___/___/___				<input type="checkbox"/> Hands are open most of the time	
Sign _____					
<b>6 months</b>	<input type="checkbox"/> Moves eyes or head in direction of sounds	<input type="checkbox"/> Eyes move well together (no squint)	<input type="checkbox"/> Laughs aloud	<input type="checkbox"/> Grasps toy in each hand	
Date ___/___/___	<input type="checkbox"/> Responds by making sounds when talked to	<input type="checkbox"/> Recognises familiar faces	<input type="checkbox"/> Uses different cries or sounds to show hunger, tiredness, discomfort	<input type="checkbox"/> Lifts head when lying on tummy	
Sign _____		<input type="checkbox"/> Looks at own hands			
<b>9 months</b>	<input type="checkbox"/> Babbles ('ma-ma', 'da-da')	<input type="checkbox"/> Eyes focus on far objects	<input type="checkbox"/> Throws, bangs toys/objects	<input type="checkbox"/> Sits without support	
Date ___/___/___	<input type="checkbox"/> Turns when called		<input type="checkbox"/> Reacts when caregiver leaves, calms when she/he returns	<input type="checkbox"/> Moves objects from hand to hand	
Sign _____					
<b>12 months</b>	<input type="checkbox"/> Uses simple gestures (e.g. lifts arms to be picked up)	<input type="checkbox"/> Looks for toy/objects that disappear	<input type="checkbox"/> Imitates gestures (e.g. clapping hands)	<input type="checkbox"/> Stands with support	
Date ___/___/___	<input type="checkbox"/> Has one meaningful word (dada, mama) although sounds may not be clear	<input type="checkbox"/> Looks closely at toys/objects and pictures	<input type="checkbox"/> Understands 'no'	<input type="checkbox"/> Picks up small objects with thumb and index finger	
Sign _____	<input type="checkbox"/> Imitates different speech sounds				

### For Health Workers...

AT EVERY VISIT: Ask the parents or caregiver if they have any specific concerns about how their child hears, sees, communicates, learns, behaves, interacts with others and uses their hands, arms, legs and body.

Tick the boxes above if the caregiver says that the child CAN do the following or if it was OBSERVED during the visit. Try to elicit the behaviour or movement if not observed through spontaneous play and interaction.

If the child can complete the task, tick the box . If the child cannot complete the task, cross the box . If you were unable to assess the task, indicate ND (not done) next to the relevant task.

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18 months	Hearing/ communication	Vision and adaptive	Cognitive/ behaviour	Motor skills	Caregiver concerns
<b>18 months</b>	<input type="checkbox"/> Understands names of at least 2 common objects e.g. cup	<input type="checkbox"/> Looks at small things and pictures	<input type="checkbox"/> Follows simple commands (e.g. 'come here')	<input type="checkbox"/> Walks alone	
Date ___/___/___	<input type="checkbox"/> Uses at least 3 words other than names			<input type="checkbox"/> Uses fingers to feed	
Sign _____					
<b>3 years</b>	<input type="checkbox"/> Child speaks in simple 3 word sentences	<input type="checkbox"/> Sees small shapes clearly at a distance (across room)	<input type="checkbox"/> Plays with other children/adults	<input type="checkbox"/> Runs well	
Date ___/___/___			<input type="checkbox"/> Uses pretend play (e.g. feeds doll)	<input type="checkbox"/> Eats on own	
Sign _____					
<b>5-6 years</b>	<input type="checkbox"/> Speaks in full sentences	<input type="checkbox"/> No reported/observed vision problems (Use Illiterate E chart if available)	<input type="checkbox"/> Interacts with children and adults	<input type="checkbox"/> Hops on one foot	
Date ___/___/___	<input type="checkbox"/> Caregiver understands child's speech		<input type="checkbox"/> Understands multiple commands (e.g. 'go to the kitchen and bring me your plate')	<input type="checkbox"/> Holds with fingers at top or middle of pencil or stick to draw	
Sign _____				<input type="checkbox"/> Dresses self	
<b>REFERRED TO:</b>	<input type="checkbox"/> Speech therapy	<input type="checkbox"/> Doctor	<input type="checkbox"/> Occupational therapist	<input type="checkbox"/> Physiotherapist	
	<input type="checkbox"/> Audiology	<input type="checkbox"/> Optometrist	<input type="checkbox"/> Doctor	<input type="checkbox"/> Occupational therapist	
	<input type="checkbox"/> Doctor	<input type="checkbox"/> Ophthalmic nurse	<input type="checkbox"/> Psychologist	<input type="checkbox"/> Doctor	
		<input type="checkbox"/> Occupational therapist	<input type="checkbox"/> Speech therapist		

If specified health professional not available, refer to one of the following health professionals for an initial developmental assessment: Doctor/physiotherapist/occupational therapist/speech therapist

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Always ask a health worker about your child's development if you are concerned about any of the following:

#### Eye problems:

- A white pupil/spot on the pupil
- Eyes are not able to fix on and follow a moving object such as a finger or toy
- One or both eyes being bigger or smaller than usual
- Crossed eyes or one eye looking in another direction

#### Hearing problems:

- Hearing loss
- Not responding to loud noises
- Seems to hear some sounds and not others
- Your child can no longer do tasks that they could before
- Your child is not communicating through speech or gestures at 18 months
- Not walking at 18 months
- Head looks large
- Head looks small
- Does not use both sides of the body/limbs equally
- Stiff arms and legs
- Floppy arms and legs

### Head Circumference

Measure every child's head circumference at 14 weeks and at 12 months. Record the child's head circumference, and refer if larger or smaller than the range shown below.

#### 14 weeks (cm)

Range 38 – 43 cm

#### 12 months (cm)

Range 43.5 – 48.5cm

### For Health Workers...

Children are at a higher risk for development problems if they have any of the following:

- Born premature/low birth weight
- Birth defect
- HIV
- Severe or moderate acute malnutrition/stunted
- Iron deficiency anaemia
- Recurrent illnesses; frequent hospitalisation

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Routine developmental surveillance recommended at every well-child visit

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Observation of child during office visit not sufficient

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Important to listen to parents and take their concerns seriously

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Asking targeted questions may help elicit concerns

# SCREENING

Defined as brief, formal, standardized evaluation

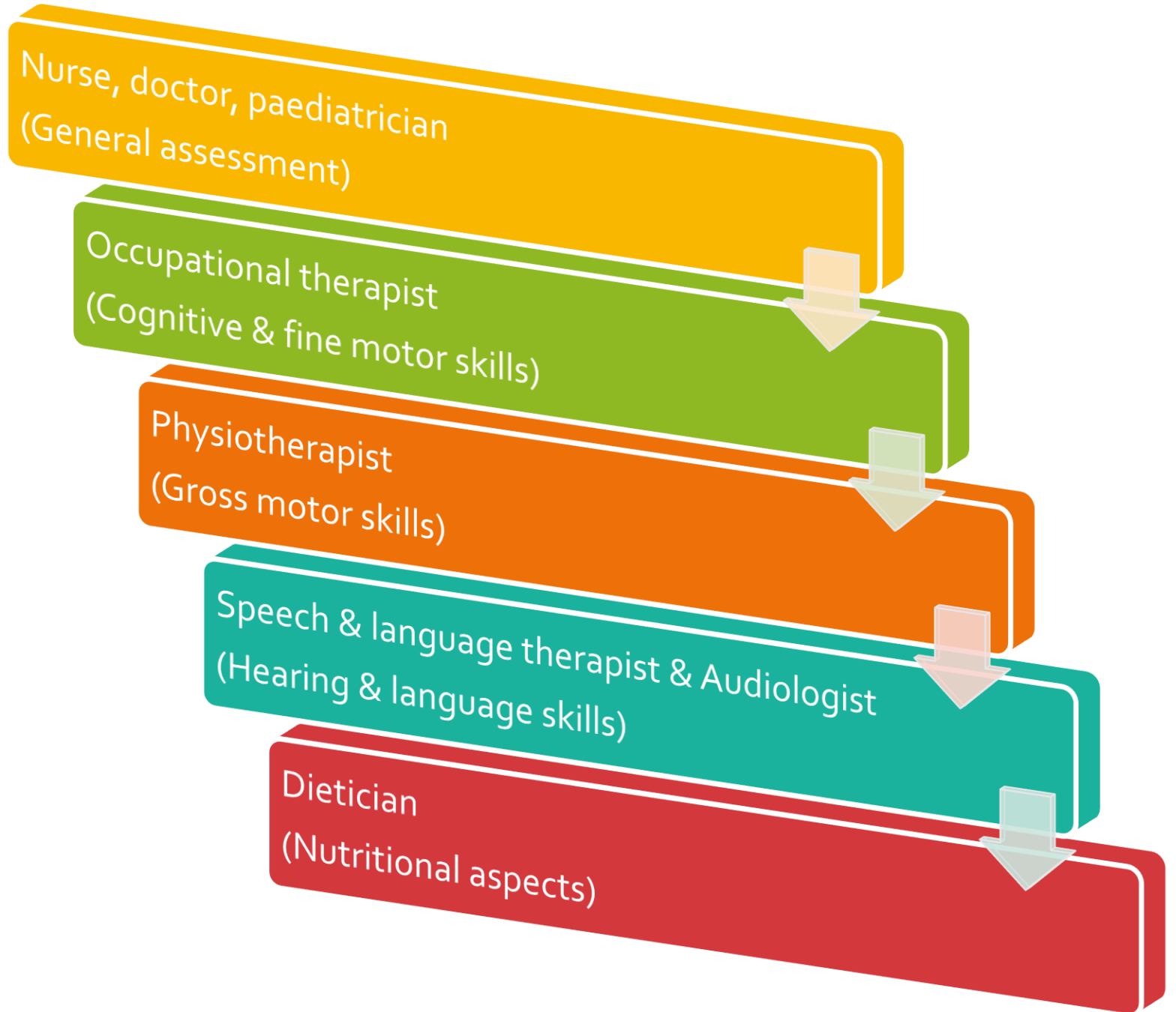
Purpose is the early identification of patients with unsuspected deviations from the norm

Screening instrument enables detection of conditions/concerns that may not be readily apparent without screening

Screening does not provide a diagnosis

Helps determine whether additional investigation is necessary

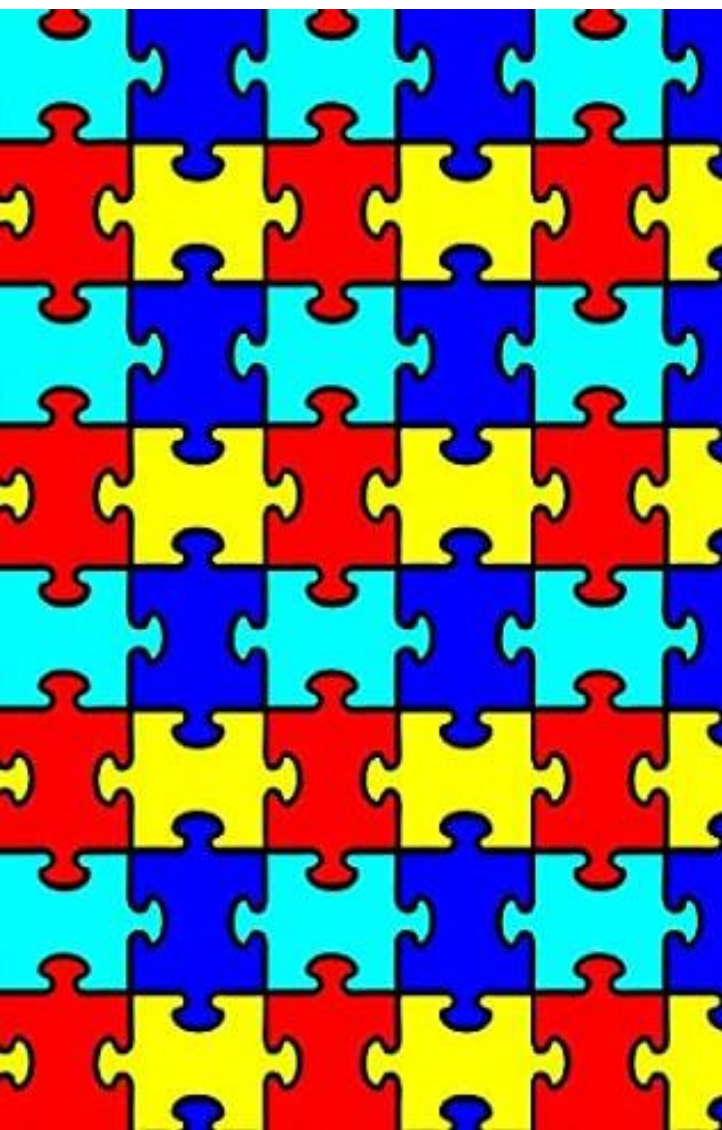
# Team members involved in screening



Benefits of  
surveillance/screening

## Early identification facilitates:

- Earlier education support
- Family support
- Delivery of appropriate medical care & Rx of associated conditions
- Genetic counselling
- Early intensive intervention



- **Screening for ASD challenging:**

- Time constraints
- Unfamiliarity with appropriate & valid ASD screening tools
- Heterogeneity in presentation of Sx & severity
- Inconsistencies in age and pattern of symptom onset
- Insufficient reimbursement



### **AAP recommends universal screening:**

- Child with any developmental concern should be screened for ASD/ referred for extensive workup
- All at 18 & 24 months

### **US Preventive Services Task Force:**

- Does not recommend universal screening
- Should screen if parental concern

### **United Kingdom (National Screening Committee):**

- Does not recommend universal screening in children < 5 yrs
- But recommend assessment of children with parental concerns (2<sup>nd</sup> screening approach)





### South African context:

Biggest income inequality in world

Diverse linguistic, cultural, ethnic & religious backgrounds



ASD screening should prioritise outreach to disadvantaged communities; consider universal screening in these community settings

Very few specialists available who can make these Ax & reach diagnostic decision

# SCREENING TOOLS

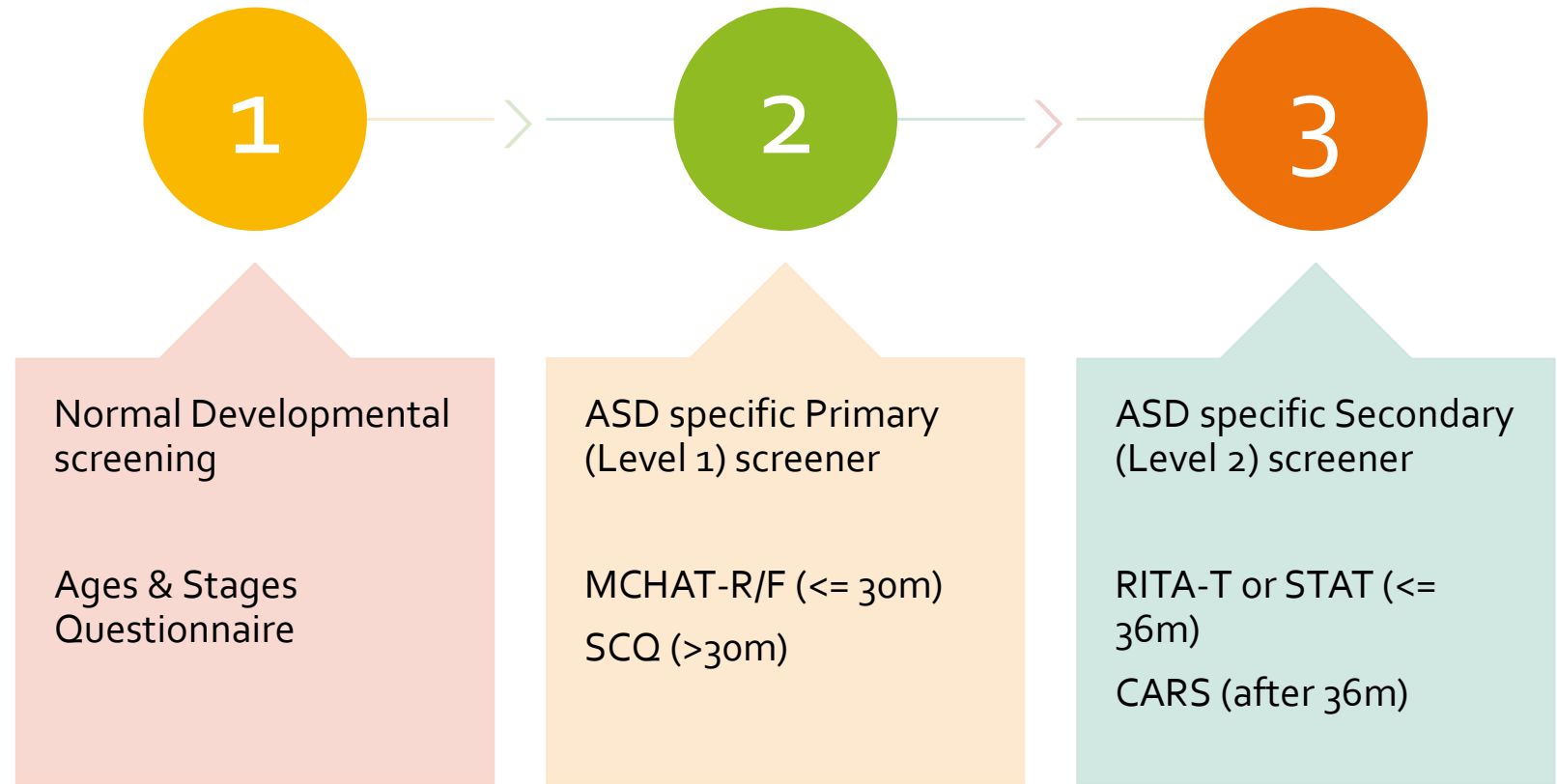
- Most developed in North America & Europe
- Scarcity of validated tools available for low- & middle-income countries (LMIC) & Africa
- Sensitivity & specificity of 90% or higher considered excellent for a medical test
- Performance of medical test in real world practice is highly dependent on many factors
  - Prevalence of the disease in the population
  - Considered in conjunction with actual population that is being assessed → positive predictive value (PPV) & negative predictive value rather than sensitivity & specificity

# A Review of Screening Tools for the Identification of Autism Spectrum Disorders and Developmental Delay in Infants and Young Children: Recommendations for Use in Low- and Middle-Income Countries

Marguerite Marlow , Chiara Servili , and Mark Tomlinson

- Identified 99 tools
  - 59 general developmental delay
  - 40 ASD
- Only 6 - ASD specific for LMIC
- Modified-Checklist for Autism in Toddlers, Revised with Follow-up (M-CHAT-R/F) → recommended tool for ASD
  - Extensively studied
  - Free to use
  - 5-10 minutes to complete

# Step-wise approach



# Research

SUPERVISOR: Dr David Griessel  
(Developmental Paediatrician)





STUDY TITLE

REFERRAL PATHWAYS OF CHILDREN DIAGNOSED  
WITH AUTISM SPECTRUM DISORDER IN  
BLOEMFONTEIN



# AIMS OF STUDY

- To investigate the referral pathways of newly diagnosed children with ASD attending the Paediatric Outpatient Departments at Universitas Academic Hospital, Pelonomi Tertiary Hospital and the private practice of a Developmental Paediatrician over a 6-month period from 1 June 2020 to 30 November 2020.
- To determine the average age of diagnosis amongst newly diagnosed children with ASD at the Universitas Academic and Pelonomi Tertiary Hospitals' Paediatric Outpatient Departments and the private practice of a Developmental Paediatrician.



# STUDY OBJECTIVES

- To determine the age of first concern regarding the development or behaviour of children diagnosed with ASD.
- To investigate the time taken from when first concern was noted until a health professional was consulted.
- To look at the action taken by the health professional with whom the child first consulted regarding the concern in development/behaviour.
- To determine the time taken to access expert services at Universitas Academic and Pelonomi Tertiary Hospitals and the private practice of a Developmental paediatrician after referral from a health professional.

- To calculate the time period from first concern until the diagnosis of ASD was made.
- To determine the average age at diagnosis of patients newly diagnosed with ASD at the Universitas Academic and Pelonomi Tertiary Hospitals' Paediatric Outpatient Departments and the private practice of a Developmental Paediatrician from 1 June to 30 November 2020.
- To describe any differences regarding the referral pathways between those patients diagnosed in the public sector (Universitas Academic and Pelonomi Tertiary Hospitals) and the private sector (Developmental Paediatrician private practice).

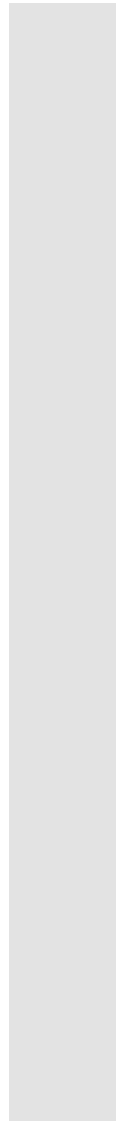
# STUDY DESIGN

- Prospective descriptive cross-sectional



# METHODOLOGY

## STUDY POPULATION

- All newly diagnosed children with ASD
    - Universitas Academic and Pelonomi Tertiary Hospitals POPD
    - Private practice of Developmental Paediatrician
- 

## INCLUSION CRITERIA

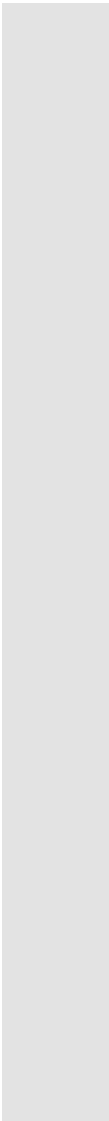
1. All newly diagnosed children with ASD irrespective of method of diagnosis used.
2. The diagnosis of ASD must be made between the period of 1 June 2020 to 30 November 2020 at the POPDs of Universitas Academic and Pelonomi Tertiary Hospitals and the private practice of a Developmental Paediatrician in Bloemfontein.
3. Children of all ages.
4. Must fulfil the DSM-5 criteria for ASD diagnosis.

# EXCLUSION CRITERIA

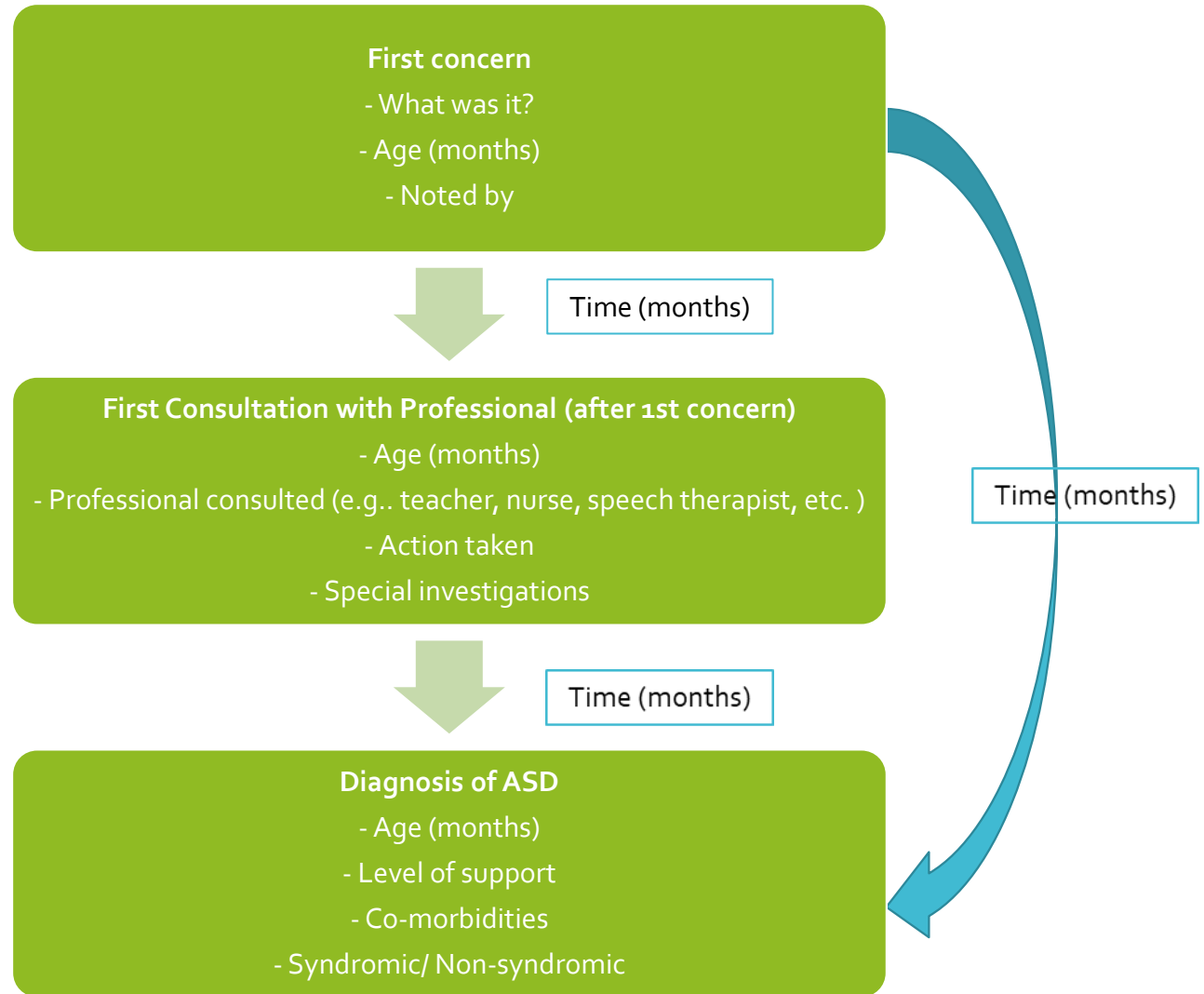
1. Children diagnosed with ASD before the commencement of the study period.
2. If assessment does not fulfil to the DSM-5 criteria for ASD diagnosis.



# DATA COLLECTION

- 
- Diagnosis of ASD
  - Informed consent from parent or caregiver
  - Completion of Data collection form
  - Captured on REDCap®
- 

# Referral pathway information collected





# RESULTS



- 39 Participants
  - Male 35 (89.7%)
  - Female 4 (10,3%)
- Place of assessment (diagnosis)
  - Universitas Academic Hospital 15 (38.5%)
  - Pelonomi Tertiary Hospital 3 (7.7%)
  - Private practice of developmentalist 21 (53.8%)

# First concern

Median age of 1<sup>st</sup> concern: 18 months

- Minimum age: 6 months
- Maximum age: 60 months

Most common 1<sup>st</sup> concerns noted:

- Speech delay
- Little or no eye contact
- Minimal reaction to verbal input/acts as though cannot hear
- Hyper- or hyposensitive to touch, taste, sight, hearing and/or smell

Noted mostly by parent/s (n=33, 84.6%)

# First consultation by professional

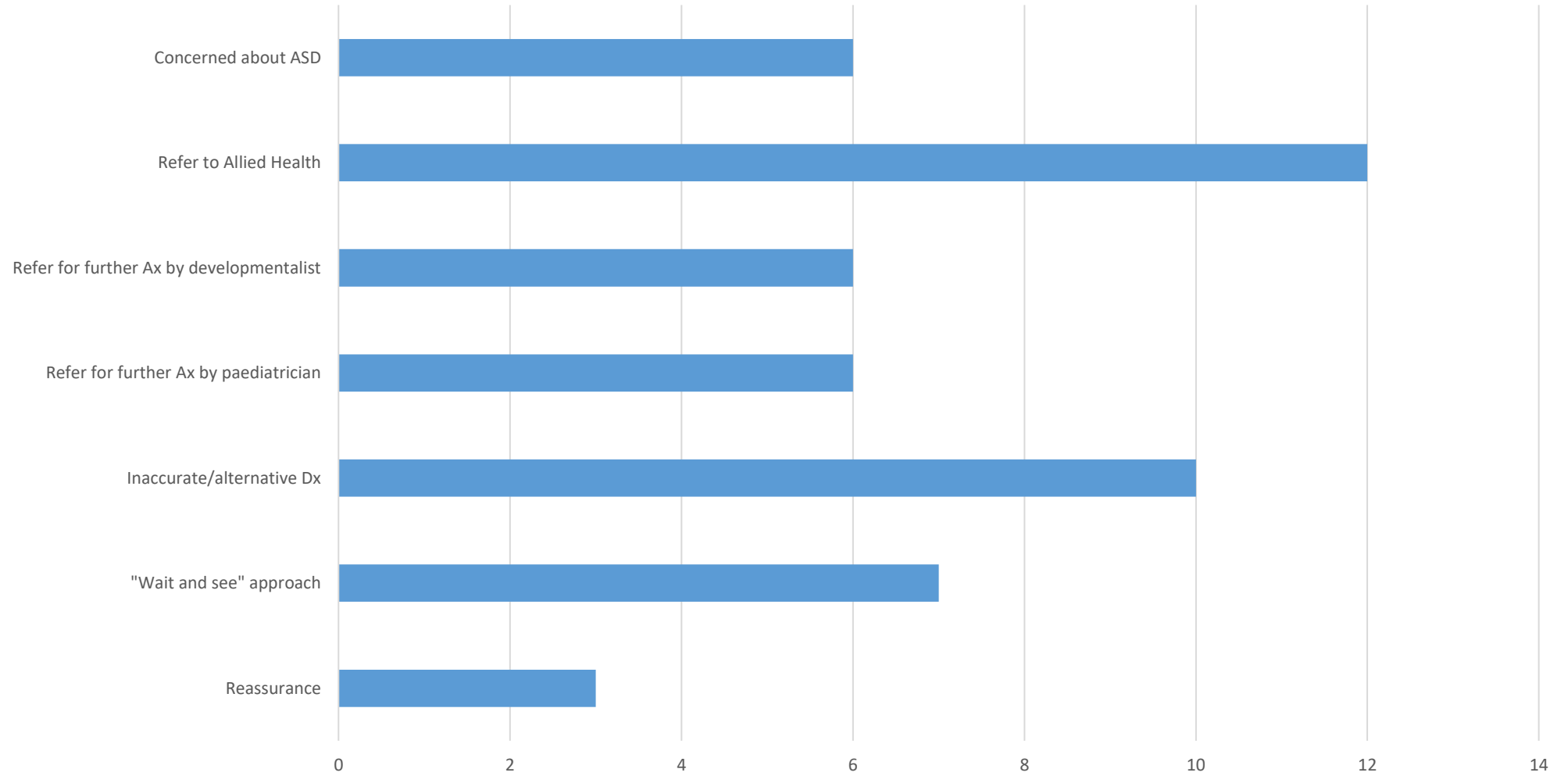
Age of 1<sup>st</sup> consultation: Median 24 months

- Minimum: 10 months
- Maximum: 70 months

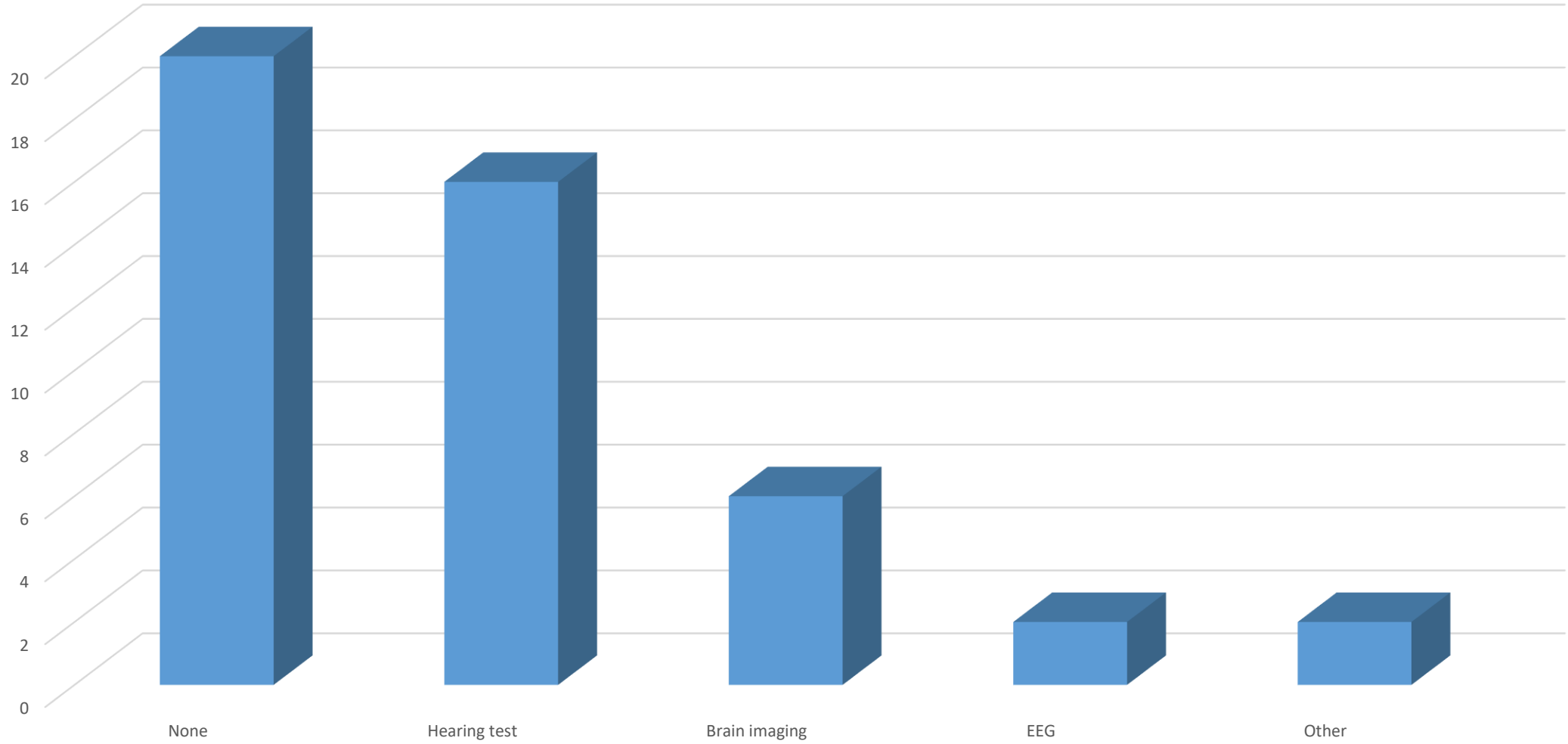
Professional consulted:

- Paediatrician
- Nurse
- Speech Therapist
- General practitioner

### Action taken by professional at first consult

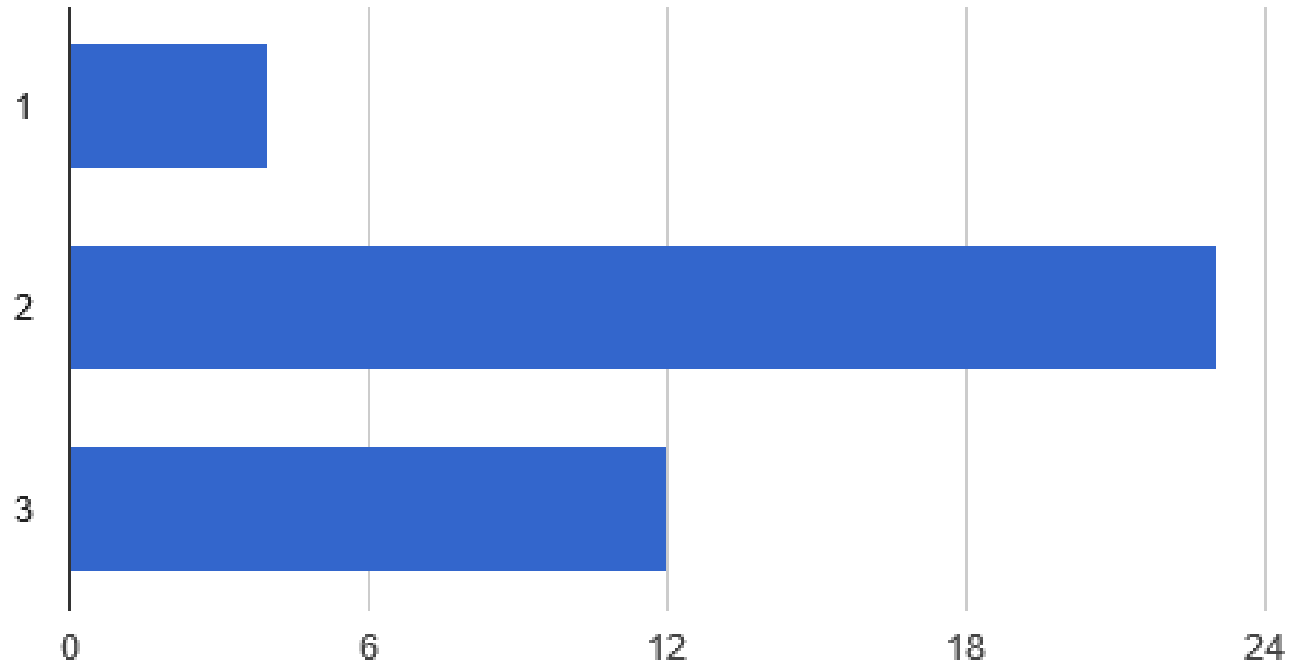


## Special Investigations



	<b>MEDIAN</b>	<b>MINIMUM</b>	<b>MAXIMUM</b>
Age at diagnosis	44.7 months	20.4 months	100.9 months
Time from 1 <sup>st</sup> professional consult to diagnosis	17 months	0.4 months	60.8 months
Time from 1 <sup>st</sup> concern to diagnosis	22.9 months	1.5 months	61.8 months

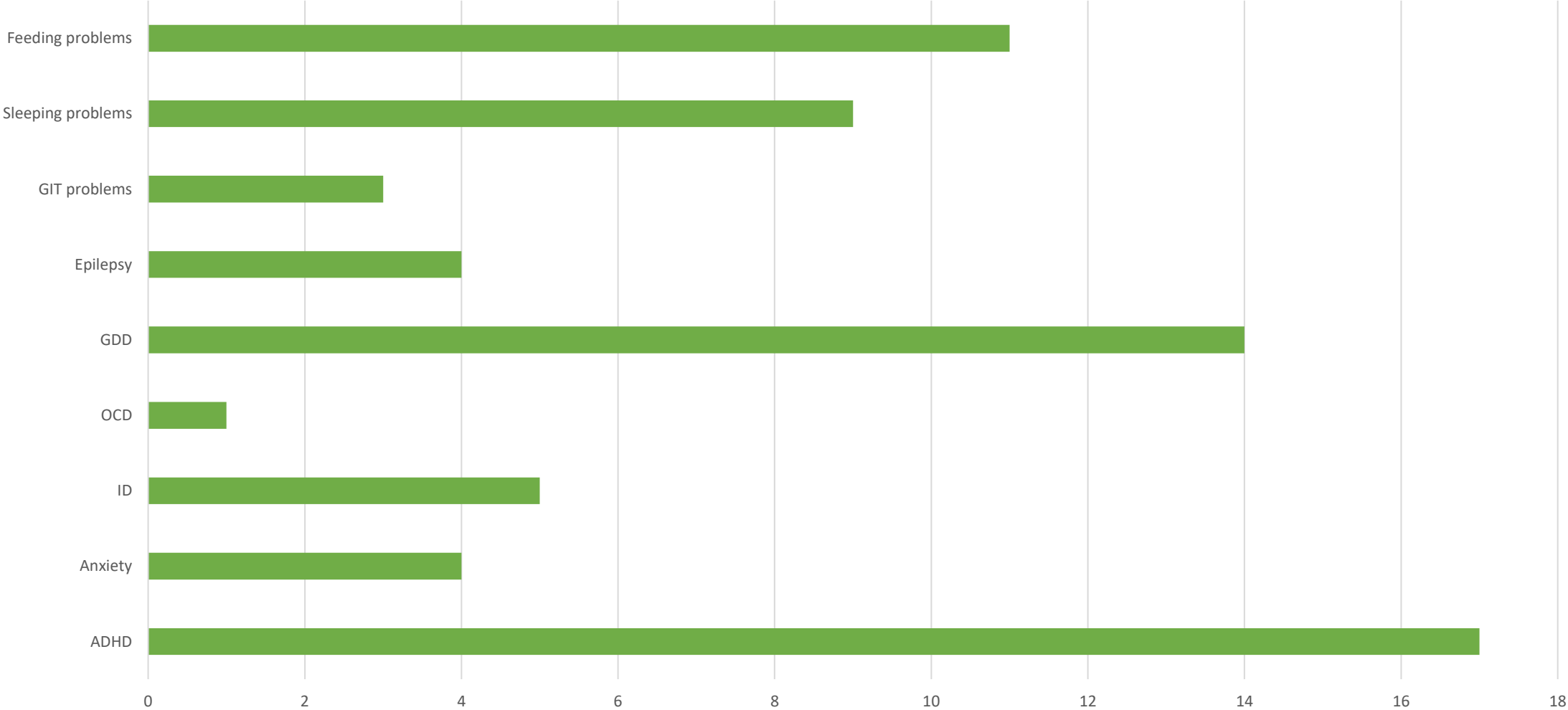
# Diagnosis



# Level of Support at Diagnosis



### Co-morbidites in children diagnosed with ASD



- Referral to alternative/ traditional healthcare workers: 4 (10.3%)
- Syndromic: 3 (7.7%)
- Family:
  - Simplex (only child) – 30 (76.9%)
  - Multiplex (other children) – 9 (23.1%)

	Public Sector (n=18)	Private Sector (n=21)
Age of Diagnosis	44.9 months	44.7 months
Age 1 <sup>st</sup> concern	18 months	22 months
Age 1 <sup>st</sup> consultation	24 months	25 months
Time 1 <sup>st</sup> consultation to diagnosis	10.7 months	19.7 months
Time 1 <sup>st</sup> concern to diagnosis	22.8 months	22.9 months

No statistical significance found between the 2 groups

# Comparison

# LIMITATIONS

- Data collection during Covid-19 lockdown period
- Small sample size
- No qualitative data collected

Still delay in the referral pathways

High functioning children missed

Co-morbidities

Not doing poorly compared to world but not doing well enough

# CONCLUSION

# Recommendations

- Further research to get qualitative data to find out why parents take long period to seek professional help
- Training of primary health care workers in the importance of using the RTHB and appropriate referrals

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