Autism Spectrum Disorders
Identification

15th Pan Arab Psychiatric Conference Cairo Sept 2018

Autism manifests in many different ways. Everyone is unique with their own abilities, talents, challenges and symptoms.

When you have met one person with autism, you have ---------

The amount of support therefore, that individual's may require can vary from constant to occasional

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What is autism as per the DSM-5

- Deficits in social communication (all 3):
  - Deficits in nonverbal communication
  - Deficits in social and emotional reciprocity
  - Deficits in maintaining relationships
- Restricted, repetitive patterns of behavior, interest, and activities (2)
  - Stereotyped motor or verbal behavior
  - Unusual sensory behavior
  - Excessive adherence to routines and ritualized bhx
  - Restricted, fixated interests
- Symptoms present in early childhood (manifest when social demands exceed capabilities)

What we know about autism: Facts and Figures

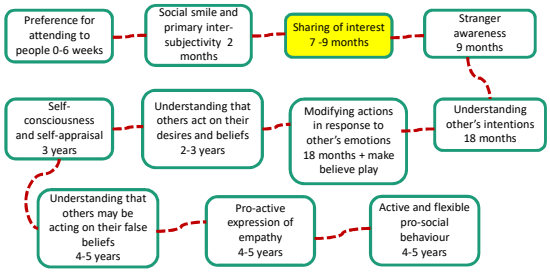
Autism affects about 1:100 (most recent 1:68)of the population

- The number of children known to have autism has increased dramatically since the 1980' s due to changes in diagnostic practice, yet many people still remain undiagnosed and without support
- It is estimated that at least 4 times more males than females are diagnosed with autism, and around two third of people with ASD also have Intellectual disability.

Social communication

- Having conversations
- Sharing our thoughts
- Sharing feelings
- Teasing and joking
- Helping
- Comforting

Outline of development of social cognition and behaviour



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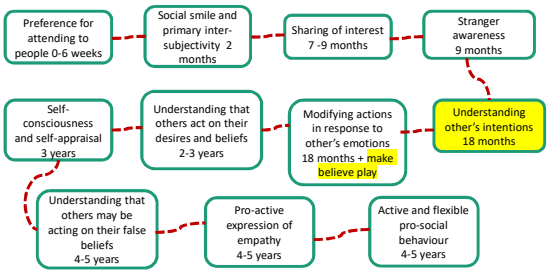

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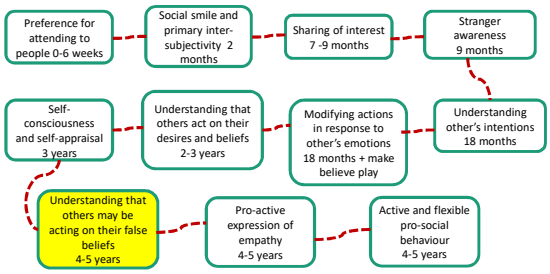
Outline of development of social cognition and behaviour



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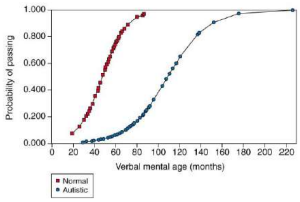
False belief understanding

Fast learning in normal development

Slow learning in autism



Autistics show 5 year delay

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What Causes ASD?

- No one cause of autism has been identified
- Most cases involve a complex and variable combination of genetic risk and environmental factors that influence early brain development

Autismspeaks.org

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Known medical causes

6-10% of children with ASD are found to have a specific medical cause

All are conditions affecting brain development:

- congenital rubella & CMV; anticonvulsants in pregnancy, foetal alcohol syndrome
- phenylketonuria, hypothyroidism
- encephalitis
- tuberous sclerosis, neurofibromatosis, West syndrome, hydrocephalus
- Fragile X syndrome, Angelman syndrome, Down syndrome, Cornelia de Lange syndrome, William's syndrome, Duchenne muscular dystrophy, non-specific chromosome abnormalities

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What does **NOT** cause autism

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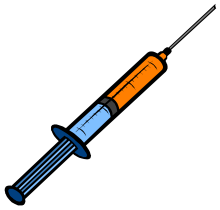
Vaccines and autism

- No reliable study has shown any link between vaccines and autism.
- Avoiding vaccines can place a child at risk of getting serious diseases



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How does autism present?

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Abnormalities in Social Interaction

- Limited interest in people
- Limited social reciprocity:
  - Social smile
  - Shared enjoyment
  - Pleasure derived from interactions
- Unusual eye contact
- Limited joint attention skills
- Poor observational/imitative learning

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Communication

- Low frequency of communication
- Limited goals of communication (instrumental versus declarative)
- Paucity of conventional and descriptive gestures (nonverbal communication)
- Stereotypical/idiosyncratic use of language (e.g., echolalia, scripting)
- Use of other's body to communicate (hand-over-hand gestures)

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Abnormalities in Play and Imagination Development

- Exploratory: present but often atypical
- Functional: may be spared but atypical
- Pretend:
  - Absent
  - Present but atypical, non-generative

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Restricted Interests and Repetitive Behaviors

- Seeking/avoiding specific visual stimuli (lights, motion, touch)
- Seeking sensory input (jumping, rocking, spinning)
- Interest in details of objects (e.g., wheels, dials)
- Hand and finger mannerisms

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Beyond the Triad of Impairments – The Sensory World of Autism

Senses play a significant role in determining our responses to a particular situation.

Many individuals with autism experience either an intensification or absence of sensory integration:

- Hyper—
- Hypo—

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Touch (includes balance and body awareness)

Hypo-

- Holding others tightly
- High pain threshold
- Self-harming (biting, gouging etc.)

Hyper-

- Finds touch painful/uncomfortable
- Sensitivity to certain clothing/textures
- Dislike of having things on hands/feet

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Autism at 6 months?

Reports of typical presentation at 6 months in children with autism

- Eye contact and affective responses to mother (Young et al., 2009)
- Social reciprocity and attention (Bryson et al., 2006; Zwaigenbaum et al., 2005)

Emerging consensus that overt symptoms of ASD begin to emerge between 6 and 12 months

Possible factors responsible for these findings:

- Natural course of autism
- Limited sensitivity of *existing* behavioral methods

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Emerging Symptoms of Autism at 12 months

- Limited response to name
  - High specificity for ASD (89%)
  - Low sensitivity (50%)
- Limited eye contact and use of communicative gestures: pointing, showing
- Delays in language: limited range and frequency of vocalizations
- Atypical behaviors: Spinning and intense visual examination of objects

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Autism changes with age

Under 5's	5+	Teens	Adults
• language and behaviour	• improved language and behaviour • may 'learn' well	• dependent, naïve • behaviour often very difficult • mental health problems	• simple repetitive life style

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Social impairment	Communication	Imagination	RRIB
Aloof	No communication	Sensory exploration	Wide range
Passive	Poor initiation	Practical use, copying	+ Routines and rituals
Active but odd	Repetitive, one sided	Repetitive and isolated	+ Routines and rituals
Overformal/ stilted	Formal, long winded	Limited pretence, own imaginary world	Intellectual interests

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Common Comorbidities

- 70% of ASD individuals have one comorbid disorder, up to 40% may have 2 or more (DSM-V)
- Medical conditions such as epilepsy and sleep problems somewhat common
- Comorbid diagnoses of ADHD, anxiety and depressive disorders, and developmental coordination disorder seen

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How is the family affected?

First degree relatives have higher incidence of major depression than the rest of the population (Bolton et al., 1998)

Relatives have 20% frequency of social phobia (Smalley et al., 1995)

- 10 times higher than controls
- Over half (64%) had first episode before the birth of autistic child

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Autism may co-exist or be a presenting feature of

- Tuberous sclerosis (Xq27.3) (16p13.3, 12q14, 9q34)
- Fragile X (Xq27.3)
- Phenylketonuria (untreated) (12q41.1)
- Rett's syndrome (X, MECP2)
- Williams' syndrome (7q 11.2)
- Turner's syndrome (XO)
- Neurofibromatosis (17q11.2)
- Angelman syndrome (Xq28, 15q11-13)

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How to identify autism?

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Levels for identification

- Population awareness and monitoring
- Screening
- Diagnostic assessment

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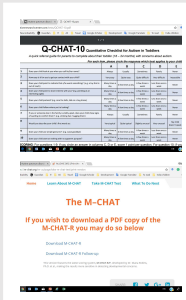

Screening

- No whole population screening recommended
- Being aware of the indicators for ASD e.g.:
  - No babbling by 12 months
  - No gesturing by 12 months
  - No single words by 16 months
  - No 2-word spontaneous phrases by 24 months
  - Any loss of any language or social skills at any age
- Screening tools may be used for children presenting with concerns about their development:
  - MCHAT: 16 to 30 months
  - Q-CHAT: 18 to 24 months

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Diagnostic tests for ASD

Autism Diagnostic Interview – Revised (ADI-R)

A standardised interviewer based questionnaire – the interviewer has to understand the concept behind each question, know the method of asking and prompting and apply the rules for coding the responses

Autism Diagnostic Observation Schedule (ADOS)

A standardised method of eliciting social and communication skills with rules for coding observations – 4 modules for different levels of verbal abilities

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What to ask? Social communication

Use of other's hand/arm as an extension of own body

Autistic speech and language abnormalities:

- Repetitive speech patterns that are odd:
  - Stereotyped content
  - Non social use
  - Out of context ritualised phrases

X common repetitions used by young children

X immediate echolalia

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What to ask? Social communication

Conversation ability	To-and-fro nature: the subject responding to what the other person says and building a dialogue on that
	The subject has to listen and respond and add

X Questions and answers

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What to ask? Social communication

- stereotyped utterances and delayed echolalia

✓ Repetitive speech patterns that are odd:
- ✓ Stereotyped content
- ✓ Non social use
- ✓ Out of context ritualised phrases

❑ Used functionally or not. The focus is on the non-social and odd use.

X common repetitions used by young children

X immediate echolalia

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What to ask? Social communication

- pronoun reversal

✓ Confusing the first person with the second or third person (code 2)

✓ I/you

✓ I/he

➢ Own name/I e.g. Brain want a biscuit: code 1

X I/me use

X Second/3rd person confusion you/he

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What to ask? Social communication

intonation/quality/rhythm

- Unusual intonation or pitch for what is being said
- Flat intonation
- Consistently abnormal volume
- Unusual rhythm

X shy low volume

X demanding loud volume

X game/family routine

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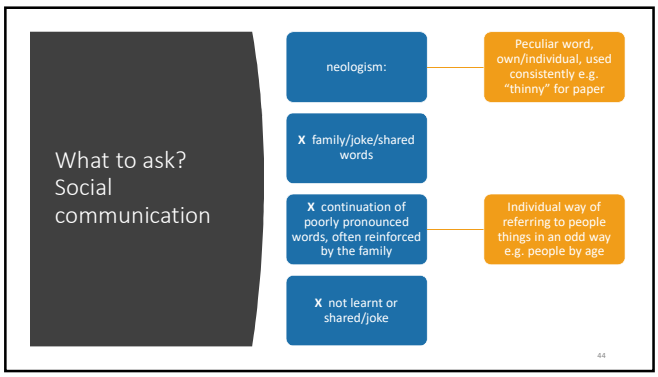

What to ask? Social interaction

Direct gaze (Eye contact)

- Social and communicative use of EC, rather than the absolute amount
- In a range of situations
- With a range of people
- Applicable up to age 5 years

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What to ask? Social interaction

pointing to express interest

- Towards an object/animal/person at a distance
- Coordinated with the eye gaze

X pointing to a picture (often a learnt response)

X pointing in response to a question

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What to ask? Social interaction

showing and directing attention

- To toys and objects in which the subject is interested
- Spontaneous
- Bringing to show items given to them or found or created by them

X to get something done with the object

X objects that are part of some special interest or preoccupation

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What to ask? Social interaction

offering to share

- Unprompted, non-routine offers to share
- May reflect the understanding of what others want or a wish to share e.g. food, toys activities (turn), offering water
- Sharing of food is difficult to interpret because it is often prompted – avoid

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What to ask? Social interaction

offering comfort

- Offering or giving comfort with a gesture, touch or vocalisation + a change in expressions to someone who is sad, ill or hurt
- Some appreciation of the person's distress

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What to ask? Social interaction

interest in children
- Watching or interacting children of same age
- Initiating interest

X interest in babies
- The quality of interaction is not coded here

<10 years age for current

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What to ask? Social interaction

Group play with peers
- Cooperative play in group situations with same age peers
- Siblings ?? +/-

X 1-1 play (dyadic)

X supervised play

< 10 years for current

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What to ask? Social interaction

Friendships
- This is about dyad, 1-1 and not groups
- Selectivity, reciprocity, mutual responsiveness, sharing

> 5 years for current

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What to ask? Social interaction

social disinhibition (knowing social cues, boundaries and rules)
- Differences in behaviour with:
  - family/friends/strangers
  - Home/public place/clinic
- Overall behaviour, not the language 9coded inn inapp statements)
- Lack of awareness
- Diff: disruptive/defiant/provocative behaviour

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What to ask? Repetitive Repetitive Behaviour and Interests (RRIB)

unusual preoccupation:
- Odd and peculiar in quality
- Unusual in intensity
- Lack of social features
- Active seeking out/talking/drawing (vs repetitive behaviours or sensory interests)
- It would be unlikely that any normally developing child form the same subculture would have an interest of that particular kind.

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What to ask? RRIB

circumscribed interest
- The content is NOT odd, but the level of interest is too intense
- The focus is too narrow
- Non-social quality
- Unusual in relation to developmental level
- Interests are clearly different from those found in normally developing children

X interests in computer games, Pokémon cards, foot ball cards,

> 3 years for current

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What to ask? RRIB

repetitive use of object or interest in parts or details

- Repetitive
- Non-functional (not the intended function)
- Unusual

X motor mannerisms

X developmentally appropriate casting, mouthing

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What to ask? RRIB

Compulsions/rituals

- Fixed sequences of behaviour that are performed 'as if' the subject feels pressure to complete them in a particular order.
- Placing objects in exact position
- Having to turn in a particular direction
- Taking a particular route
- Having to turn off a particular light/close a particular door first
- Diff: difficulties with minor change – others have made the change; repetitive use of objects is non-functional

X bedtime routines

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What to ask? RRIB

- Unusual sensory interest
- Unusually strong seeking of stimulation of sight/touch/smell/taste/sound

X smelling food

X developmentally appropriate mouthing

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What to ask? RRIB

undue general sensitivity to noise

Increased sensitivity to everyday sounds

= some behavioural change e.g. covering of ears

X reaction to sudden loud sound

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What to ask? RRIB

difficulties with minor change with own routines or personal environment

- Changes in where or how the subject carries out daily activities
- Minor aspects that would not be upsetting to most children

X excludes reaction to major changes e.g. house move

X excludes normal distress shown at the change of family routines – it is the minor aspect that would not be upsetting for most that is coded here.

Diff: compulsions re initiated by the subject and changes to the environment by others

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What to ask? RRIB

unusual attachment to object

- The object must be unusual e.g. clothes peg, Lego piece

X cuddly toys, blanket, doll,

A specific object NOT a type of object e.g. a particular peg not any peg

Distress or insistence on not being able to have the object to carry around

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What to ask? RRIB

complex mannerisms or stereotyped body movements

- Spinning
- Bouncing up and down
- Repetitive leaning forward with hand waving

X rocking (unless part of a complex movement)

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Play

Pretend or imaginative play

- **Creative** and **varied** use of actions and objects in play
- A story or a sequence
- Use of doll/animal/toy as an agent

X taught sequence

X repetitive sequence

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Play

imaginative play with peers

- Play may be simple or complex, but
  - Has to be socially interactive
  - Varied
  - With 1 or more children
  - The subject takes lead as well as follows
  - Play with siblings Ok as long as the above criteria are present

X the well practised family routines

- 4-10 years (such play happens in the middle childhood)

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What to observe?

Social skills

- Asking
- Showing
- Initiating an interaction
- Responding to interaction
- Communication, conversation
- Reciprocity
- Sharing enjoyment
- Play and social imagination
- Peer interaction and friendships
- Understanding of social boundaries

Features

- Eye contact
- Gestures and facial expressions
- Odd intonation/delayed echolalia/stereotypical phrases
- Strong/unusual interests
- Compulsions/repetitive behaviour
- Unusual sensory interest/ hypersensitivity
- Unusual movements
- Social anxiety
- Attention/hyperactivity

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How to observe?

School observation (or ask the school): Interest in other children, friends, social behaviour

Clinic observation:

- Free play
- Joint play
- Joint attention
- Generating interest and using time delay or pause (to initiate asking)
- Drawing attention or asking to show (for joint attention)
- Social games (for social initiation and response)
- Conversation
- Interview (to ask about social understanding, feelings, difficulties)
- Story reading and story making

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Diagnostic cautions – missing the diagnosis

- Signs and symptoms will not always have been recognised by parents
- Signs or symptoms may have previously been masked by the child or young person's coping mechanisms and/or a supportive environment
- Be aware, but don't make assumptions about language delay, behaviour difficulties or disruptive home experiences as the reason for the signs
- Autism may be missed in children or young people with a learning (intellectual) disability and in those who are verbally able
- Autism may be under-diagnosed in girls

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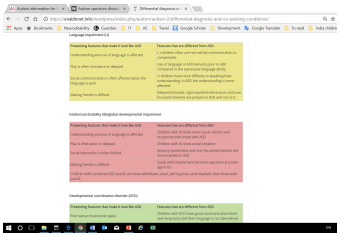
Diagnostic cautions – over diagnosing

- Some other neuro-developmental and mental health conditions have features that are similar to those seen in ASD.
- See www.enablenet.info differential diagnosis.
- To diagnose ASD one has to know about these other conditions too, not just about autism.
- Otherwise, "if the only tool you have is a hammer, everything looks like a nail"!



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Functional and other difficulties

- feeding problems, including restricted diets
- urinary incontinence or enuresis
- constipation, altered bowel habit,
- sleep disturbances
- vision or hearing impairment

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Essential physical examination

- Growth: height and weight
- skin stigmata of neurofibromatosis or tuberous sclerosis using a Wood's light
- signs of injury, for example self-harm or child maltreatment
- congenital anomalies and dysmorphic features including macrocephaly or microcephaly

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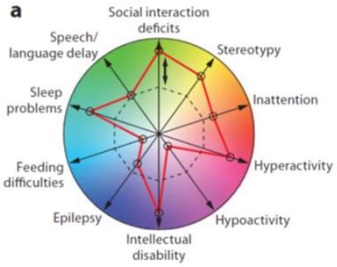

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Investigations

Do not routinely perform any medical investigations as part of an autism diagnostic assessment, but consider the following in individual circumstances and based on physical examination, clinical judgment and the child or young person's profile:

- genetic tests, if there are specific dysmorphic features, congenital anomalies and/or evidence of a learning (intellectual) disability
- electroencephalography if there is suspicion of epilepsy

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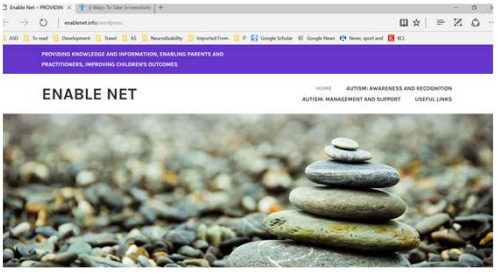
For further information: go to ENABLENET.INFO



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