WOLLO UNIVERSITY

COLLEGE OF MEDICINE AND HEALTH SCIENCE

DEPARTMENT OF COMPREHENSIVE NURSING

Assignment on :pediatric nursing

Acknowledgment

First I would like to thanks our techer mulugeta W./ass.prof/ to give our group assignment and allow presentation.

Out line

1. Down syndrome

- Definition
- Cause
- Pathophysiology
- Sing and symptoms
- Diagnosis
- Management
- complication



Down Syndrome

1. DOWN'S SYNDROME/ TRISIOMY 21

1.1 DEFINITION

Down's syndrome is a chromosomal abnormality involving an extra chromosome characterized by

- a typical physical appearance and
- mental handicap.

Down syndrome is a chromosomal condition that is associated with intellectual disability, but the intellectual disability is usually mild to moderate.

1.2 CAUSES

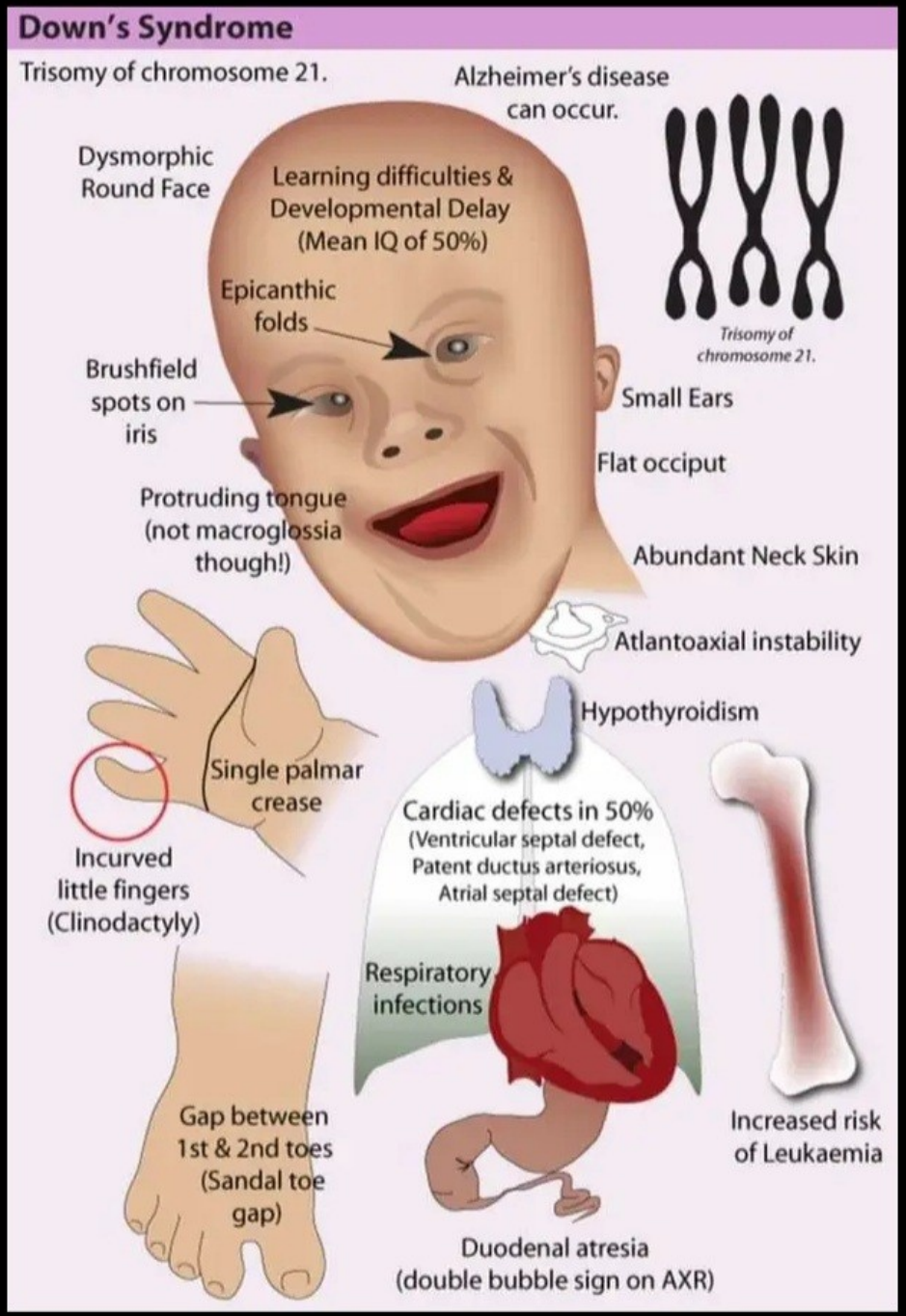
- Most cases (95%) result from trisomy 21, which means each cell in the body has three copies of chromosome 21 instead of the usual two copies.
- Less commonly, it occurs when part of chromosome 21 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) in a parent or very early in fetal development.

1.3 SIGN AND SYMPOM

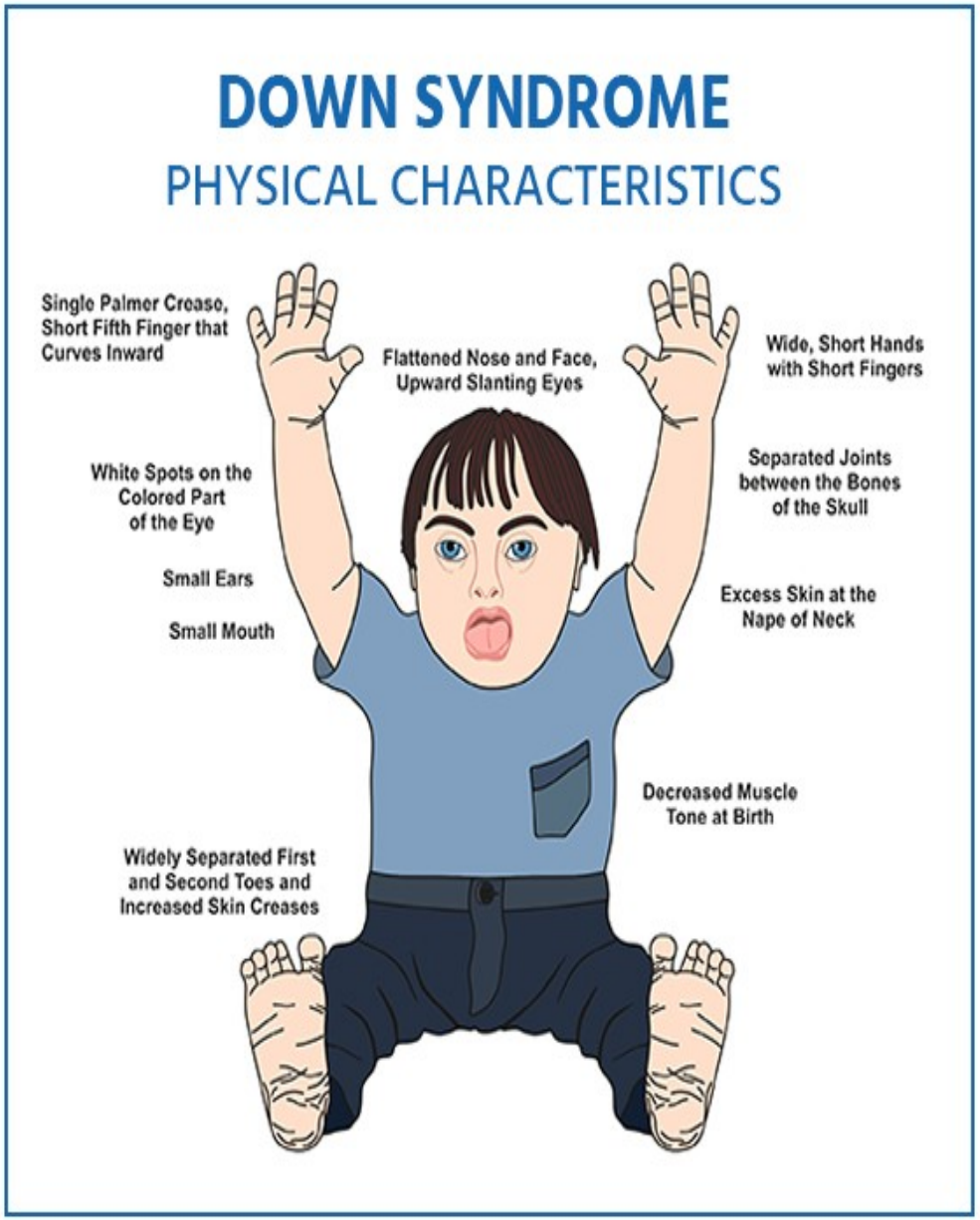
Typical craniofacial appearance

- microcephaly
- Round face and flat nasal bridge
- Upslanted palpebral fissures
- Epicanthic folds (a fold of skin running across the inner edge of the palpebral fissure)
- Brush field spots in iris (pigmented spots)
- Small mouth and protruding tongue
- Small ears
- Flat occiput and third fontanelle

Down's Syndrome



Con..


DOWN SYNDROME
PHYSICAL CHARACTERISTICS
Single Palmer Crease, Short Fifth Finger that Curves Inward
Flattened Nose and Face, Upward Slanting Eyes
Wide, Short Hands with Short Fingers
White Spots on the Colored Part of the Eye
Separated Joints between the Bones of the Skull
Small Ears
Excess Skin at the Nape of Neck
Small Mouth
Decreased Muscle Tone at Birth
Widely Separated First and Second Toes and Increased Skin Creases

Other s/s

CENTRAL NERVOUS SYSTEM

- Hypotonia
- Developmental delay
- Poor Moro reflex
- Small dysplastic ears

CARDIOVASCULAR

- Endocardial Cushing defects
- Ventricular septal defect
- Atrial septal defect
- Patent ductus arteriosus
- Pulmonary hypertension

MUSCULOSKELETAL

- Joint hyper flexibility
- Short neck, redundant skin
- Short metacarpals and phalanges
- Single transverse palmar creases
- Wide gap between 1st and 2nd toes
- Pelvic dysplasia
- Short sternum
- Two sternal manubrium ossification centers

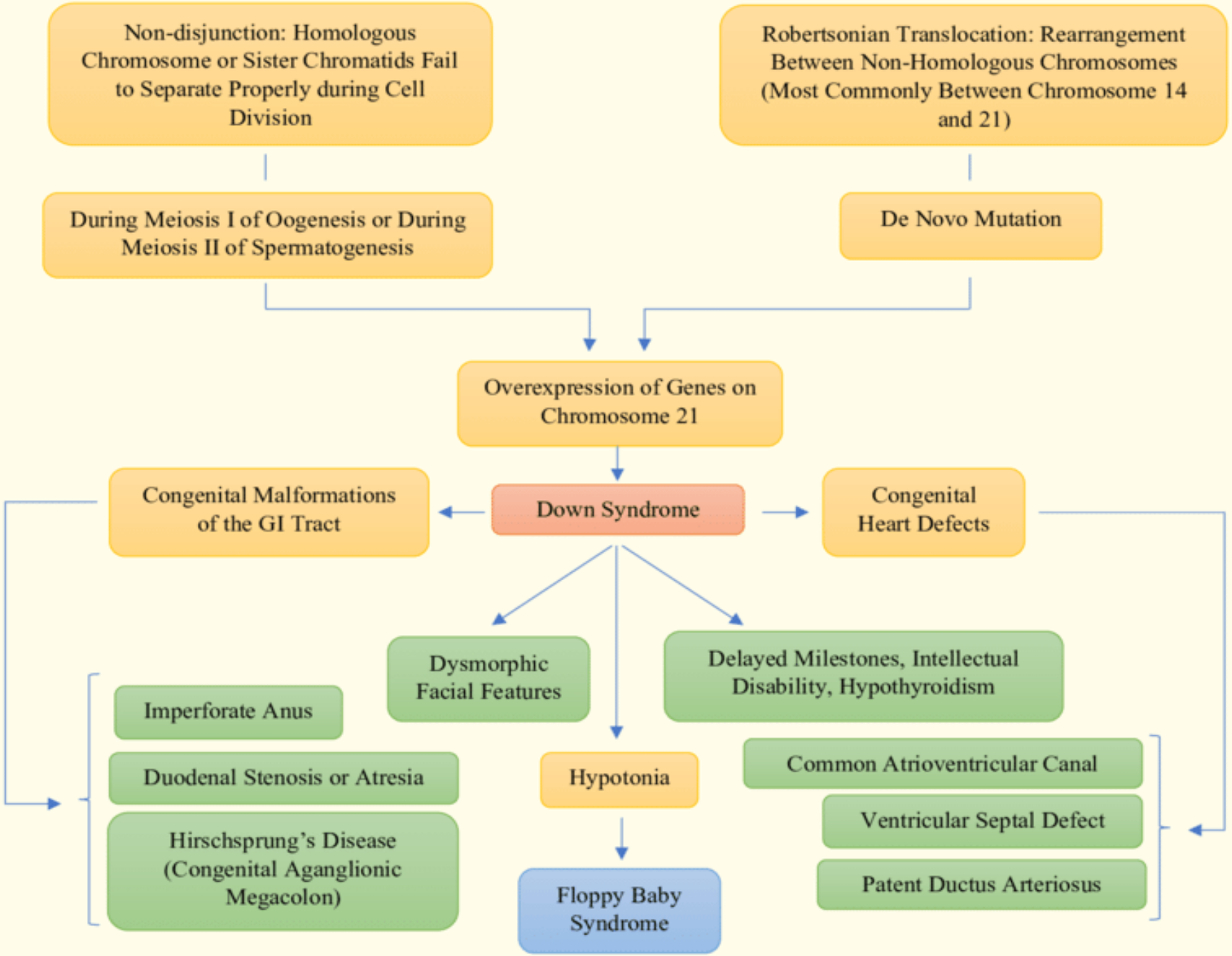
GASTROINTESTINAL

- Duodenal atresia
- Trache oesophageal fistula
- Hirschsprung disease
- Imperforate anus

1.4 PATHOPHYSIOLOGY

Down syndrome is usually caused by an error in cell division called nondisjunction

- During meiosis, one pair dose not dived and the whole pair goes to one daughter cell.
- In the resulting cell one will have 24 chromosomes and the other will have 22
- So if a sperm or egg with abnormal of chromosomes merges with a normal mate. The resulting fertilized egg will have an abnormal # of chromosomes.
- In Trisomy 21, one cell has two 21st chromosomes instead of one, so the resulting fertilized egg has three 21st chromosomes


Non-disjunction: Homologous Chromosome or Sister Chromatids Fail to Separate Properly during Cell Division
Robertsonian Translocation: Rearrangement Between Non-Homologous Chromosomes (Most Commonly Between Chromosome 14 and 21)
During Meiosis I of Oogenesis or During Meiosis II of Spermatogenesis
De Novo Mutation
Overexpression of Genes on Chromosome 21
Congenital Malformations of the GI Tract
Down Syndrome
Congenital Heart Defects
Dysmorphic Facial Features
Delayed Milestones, Intellectual Disability, Hypothyroidism
Imperforate Anus
Duodenal Stenosis or Atresia
Hirschsprung's Disease (Congenital Aganglionic Megacolon)
Hypotonia
Floppy Baby Syndrome
Common Atrioventricular Canal
Ventricular Septal Defect
Patent Ductus Arteriosus

1.5 DIAGNOSTIC TESTS

- ***Chorionic Villus Sampling /CVS***/ :-sample of placenta taken either through the cervix or through a needle inserted the abdomen. Test can be performed 8 and 12 weeks
- ***Amniocentesi:-s*** removal a small amount of amniotic fluid through a needle inserted in the abdomen. Cells are analyzed for the presence of chromosomal abnormalities. Test performed between 12 and 20 weeks of pregnancy
- ***Percutaneous Umbilical Blood Sampling/PUBS/*** :- uses a needle to retrieve a small sample of blood from the umbilical cord. Usually performed after 20 weeks

Con

Karyotype:- After the baby is born, if Down syndrome is suspected a blood or tissue sample stained to show chromosomes grouped by size,#, and shape can be performed to confirm the diagnosis

General appearance:- of the neonate

*Confirmation is established by chromosomal analysis (karyotype) using bone marrow aspiration or leucocytes culture.

1.6 MANAGEMENT

Treatment for Down syndrome

1. Early Intervention and Educational Therapy

- There are special type of educators, speech therapist, occupational therapist and other workers .

2. Physical Therapy

- Physical therapy is even more crucial than educational therapy in many aspects because physical disability increases over the period of time.

Physical therapy can improve

- The ability to turn around
- Ability to crawl in infants
- Ability to walk in children
- Improve level of interaction

3. Speech-language therapy

4. Occupational Therapy

- This therapy is mostly used in functions related to everyday life.
- It includes-
- self-care skills such as eating, getting dressed and writing, etc.
- Special tools are used to improve everyday activities and functions

5. Emotional and Behavioral therapy

- It makes people more understanding of the condition or situation that helps them to improve behavior and undesired levels of frustration. Children with Down syndrome may also be vulnerable to Attention Deficit Hyperactivity Syndrome.

1.7 COMPLICATIONS

- Delayed language, social, & motor development
- Growth retardation
- Congenital heart disease (50%)
- Atrial or Ventricular septal defect
- Respiratory infections, Pneumonia
- Vision problems, Cataracts

Con..

- Hearing loss (> 60%)
- GI blockages/problems, esophageal atresia, duodenal atresia, Celiac disease
- Thyroid dysfunctions
- Sleep apnea (50%-75%)
- Increased risk for Dementia
- Higher risk for ALL (Acute Lymphocytic Leukemia)- (1/150)

Thank you



Out line

2. Fragil x syndrom

- Definition
- Cause
- Pathophysiology
- Sing and symptoms
- Diagnosis
- Management
- complication


Fragile X Syndrome

2. FRAGILE X SYNDROME

2.1 DEFINITION

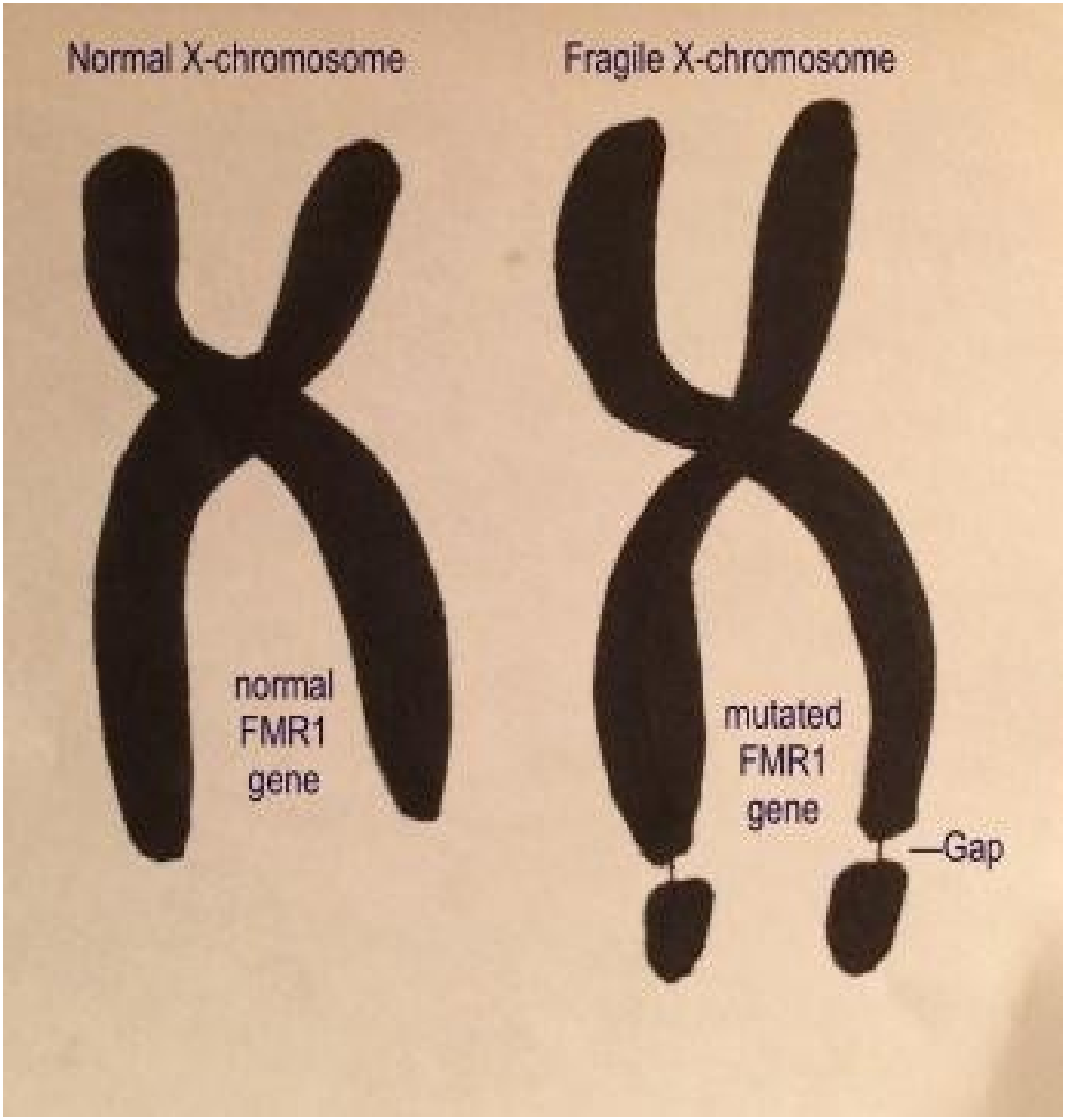
Fragile x syndrome / FXS / is a genetic disorder caused by change in a gene called fragile x messenger ribonucleoprotien 1/FMR1/.FMR1 usually makes a protein called fragile x messenger ribonucleoproteion /FMRP/.this protein is needed for brain development

con

- FXS affects both male and females.
- Females often have milder symptoms than males.

2.2 CAUSE

- ❖ Mutations in the FMR1 gene cause Fragile X syndrome
- ❖ caused by a mutation in which a DNA segment, known as the CGG triplet repeat
- ✓ Normal CGG:- repeat from 5 to 44 repeats
- ✓ Intermediate:- 45 to 54 repeats
- ✓ Permutation :-55 to 200
- ✓ Full mutation/ FXS /:- more than 200 repeats


Normal X-chromosome
Fragile X-chromosome
normal
FMR1
gene
mutated
FMR1
gene
Gap

2.3 SIGNS AND SYMPTOMS

- Intellectual disabilities
- Developmental delays/not sitting, walking, talking at the same time as other children the same age /
- Social and behavior problems /such as not making eye contact, anxiety, acting and speaking with out thinking /

Fragile X syndrome:

Common Physical Features

Prominent, Broad Forehead

Large Ears

Long Face

Strabismus (Squint)

Prominent Jaw, Dental Crowding High Arched Palate



Murmur/ Mitral Valve Prolapse

Hollow Chest

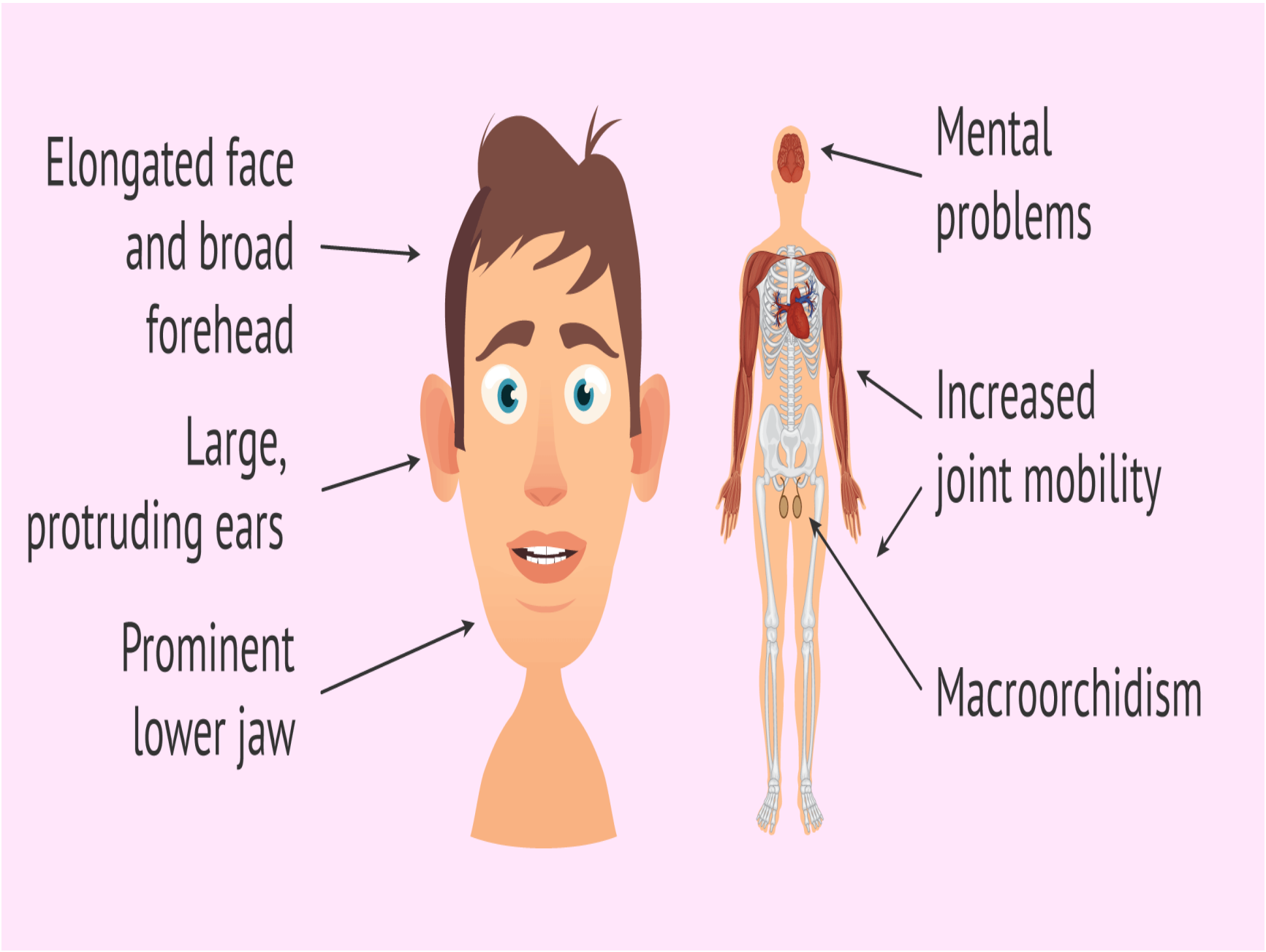
Hypotonia / Joint Laxity

Scoliosis

Macro-Orchidism

Symptoms

- Autism Spectrum Disorders
- Intellectual Disability
- Distinct facial features

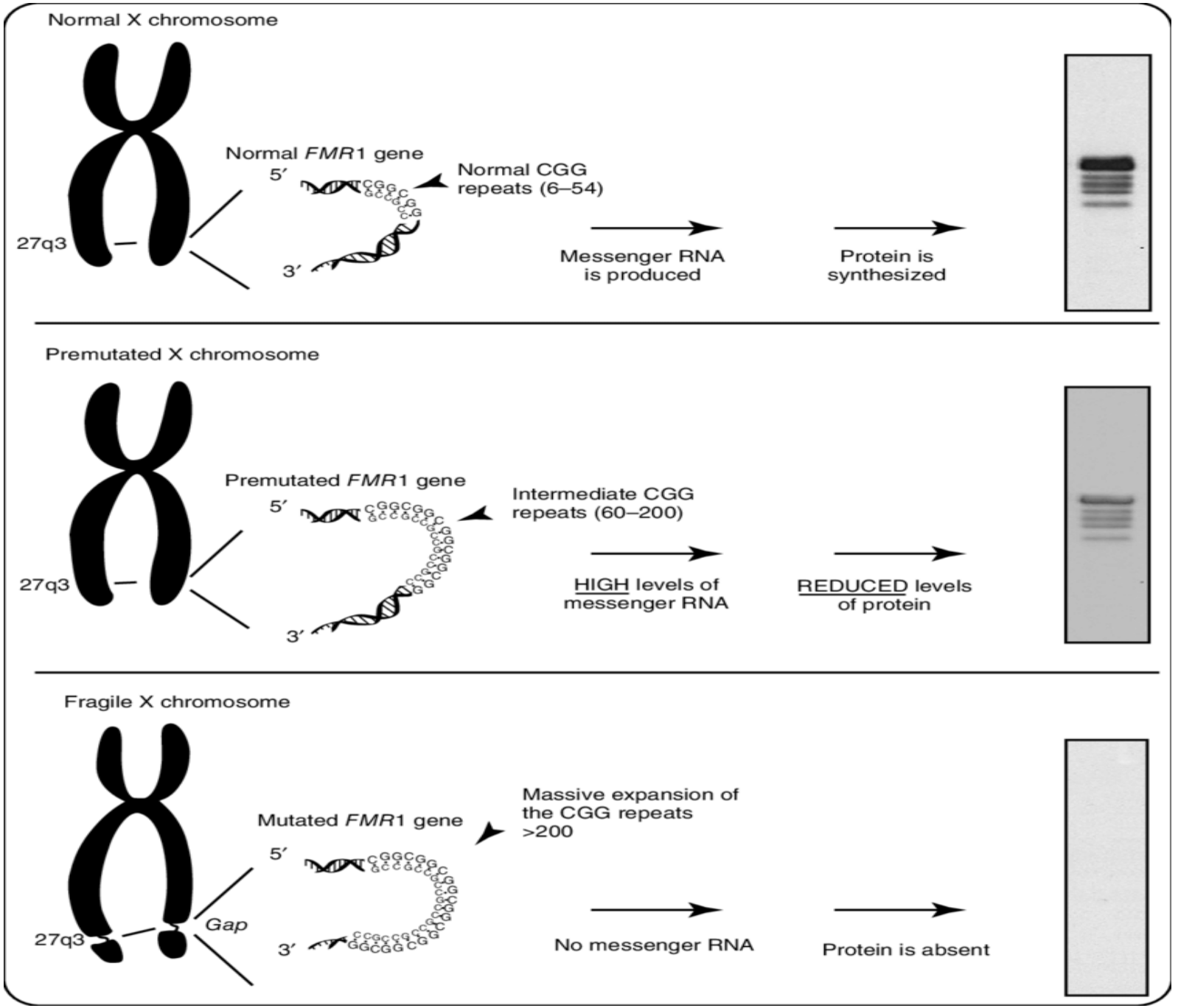

Elongated face and broad forehead
Large, protruding ears
Prominent lower jaw
Mental problems
Increased joint mobility
Macroorchidism

2.4 PATHOPHYSIOLOGY

- It is develops by the full mutation (FM) of the FMR1 gene, which is characterized by the excessive expansion of CGG trinucleotide repeats (gerater than 200)
- CGG triplet repeats are hypermethylated with consequent transcriptional gene silencing, halting gene expression, thereby resulting in a reduction or absence of FMRP.

Con..

- The number of CGG trinucleotide repeats expands with each subsequent generation, growing from pre mutation in women (PM; 55–200 repeats) to a FM in their offspring.
- For PM alleles with more than 99 CGG repeats, the risk of transition from PM to FM approaches 100%.


Normal X chromosome
27q3
Normal FMR1 gene
5′
3′
Normal CGG repeats (6–54)
Messenger RNA is produced
Protein is synthesized
Premutated X chromosome
27q3
Premutated FMR1 gene
5′
3′
Intermediate CGG repeats (60–200)
HIGH levels of messenger RNA
REDUCED levels of protein
Fragile X chromosome
27q3
Gap
Mutated FMR1 gene
5′
3′
Massive expansion of the CGG repeats >200
No messenger RNA
Protein is absent

2.5 DIAGNOSIS

- The FMR1 DNA Test (sometimes called the Fragile X DNA Test)
- DNA testing detects more than 99% of individuals (both males and females) with Fragile X syndrome, as well as Fragile X carriers.

con

- There are three general circumstances in which Fragile X testing should be considered:

1. Clinical symptoms that suggest Fragile X Syndrome, fragile x associated tremor/ataxia syndrome /FXTAS/, or Infertility/ Fragile X Primary Ovarian Insufficiency/FXPOI/.
2. A family history of Fragile X syndrome, FXTAS, intellectual or learning disabilities or autism of unknown cause, or infertility.
3. Family or personal history of a Fragile X genetics and inheritance (i.e., carrier).

2.6 TREATMENT

- **Non-Pharmacological Therapy of FXS**
- no cure for Fragile X syndrome
- Given the proper education, therapy, and support
- Supportive therapy for children who have Fragile X syndrome includes:

- Occupational Therapy/OT/
- Speech and language Therapy/SLT/
- Physical Therapy /PT/
- Cognitive Behavioral Therapy/CBT/
- Music Therapy
- Exercise
- Applied Behavior Therapy/ABT/

Psychopharmacology in FXS

The most common psychiatric symptoms that are sought to improve with drug treatment in FXS are:

- ADHD, anxiety
- Aggressiveness
- sleep problems

Thank you



3.Out line

3. Autism syndrom

- Definition
- Cause
- Pathophysiology
- Sing and symptoms
- Diagnosis
- Management
- complication


AUTISM

3. AUTISM SPECTRUM DISORDER (ASD)

3.1 DEFINITION

- Autism spectrum disorder (ASD) is a developmental disorder that affects communication and behavior.

Con..

- According to the *Diagnostic and Statistical Manual of Mental Disorders (DSM-5)*, a guide created by the American Psychiatric Association used to diagnose mental disorders, people with ASD have:

❖ Difficulty with communication and interaction with other people

❖ Restricted interests and repetitive behaviors

❖ Symptoms that affect the person's ability to function in school, work, and other areas of life

3.2 CAUSES

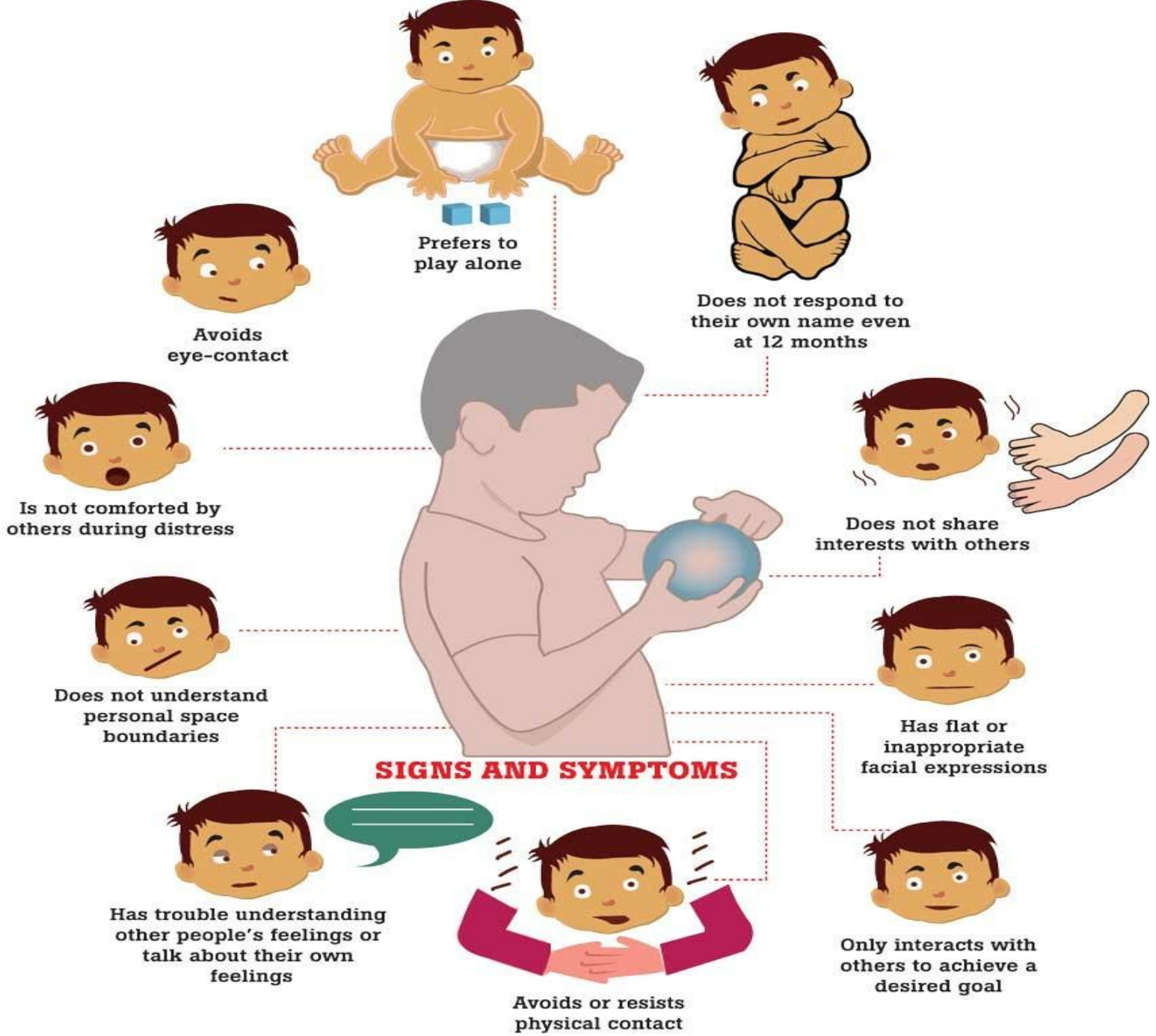
- The exact causes of ASD, unknown

some factors that increase the risk of developing ASD include:

- Having a sibling with ASD
- Having older parents
- Having certain genetic conditions

3.3 SIGNS AND SYMPTOMS OF ASD

- have difficulty with social communication and interaction and have restricted interests and repetitive behaviors



©DM GraphicDesk

MINDJOURNAL

SIGNS OF AUTISM



Unique or abnormal speech



Abnormal behaviors in social situations



Repetitive, limited behavior patterns



Narrow interests



Lack of empathy or insensitive



Sensory sensitivities



Repetitive, abnormal movements



Preference for predictable routines



Sleep problems

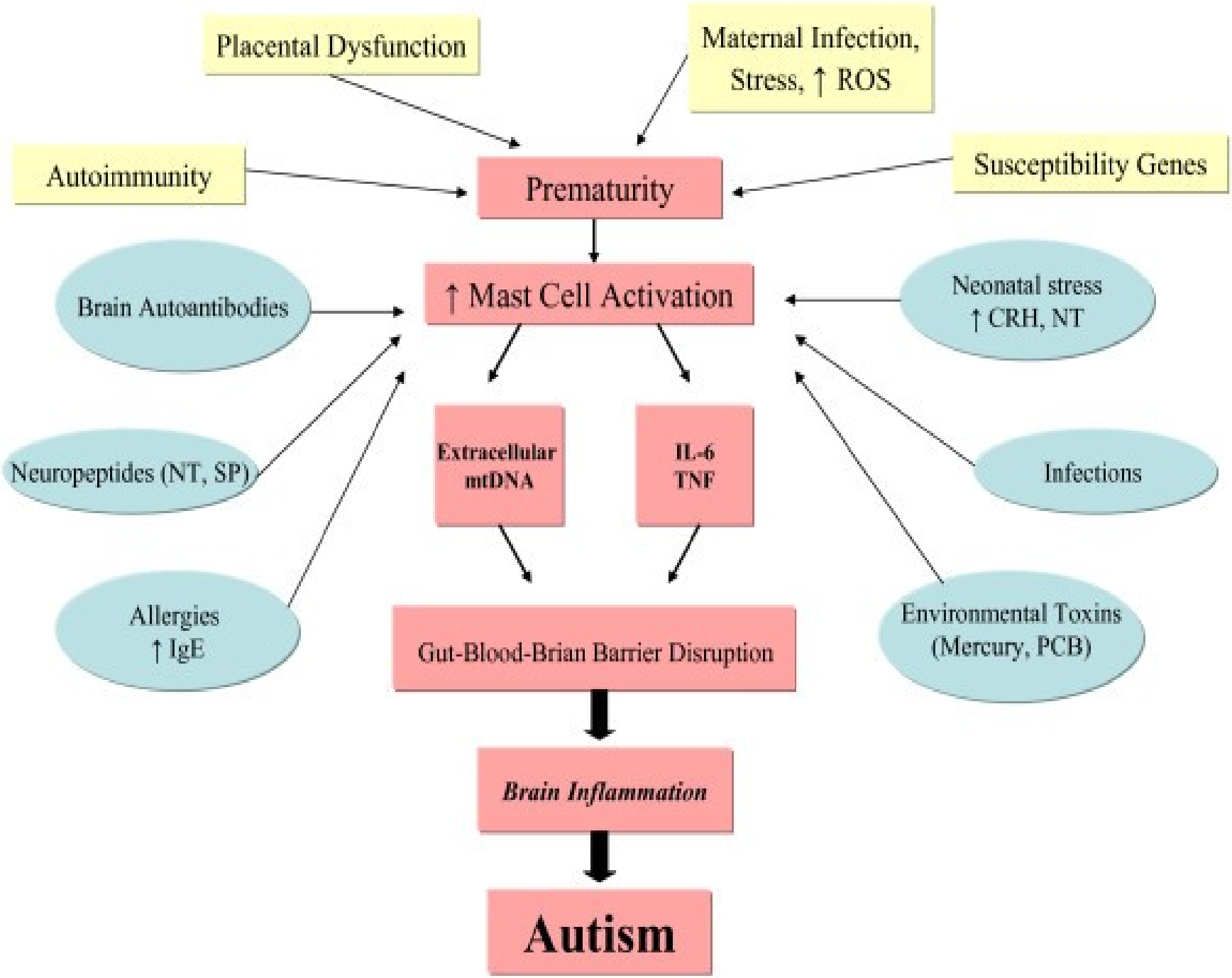


Anxiety & depression

MIND HELP

3.4 PATHOPHYSIOLOGY

Autism appears to result from developmental factors that affect many or all functional brain systems, and leads to disruption in the development of the brain.


Placental Dysfunction
Maternal Infection, Stress, ↑ ROS
Autoimmunity
Prematurity
Susceptibility Genes
Brain Autoantibodies
↑ Mast Cell Activation
Neonatal stress ↑ CRH, NT
Extracellular mtDNA
IL-6 TNF
Neuropeptides (NT, SP)
Infections
Allergies ↑ IgE
Gut-Blood-Brian Barrier Disruption
Environmental Toxins (Mercury, PCB)
Brain Inflammation
Autism

Diagrammatic representation of proposed events and interactions during the perinatal period that may contribute to autism. Placental dysfunction, as well as autoimmunity, maternal infection and gestational stress lead to prematurity. Defective neuronal development and susceptibility genes make the infant vulnerable to environmental triggers that activate mast cells to release mediators that disrupt the gut-blood–brain barriers causing brain inflammation. CRH, corticotropin-releasing hormone; IgE, immunoglobulin E; IL, interleukin; LPS, lipopolysaccharide; MCP-1, macrophage chemo-attractant protein-1; mtDNA, mitochondrial DNA; NT, neurotensin; PCB, polychlorinated biphenyl; ROS, reactive oxygen species; SP, substance P; TNF, tumor necrosis factor.

Advertisement

3.5 DIAGNOSE

There are certain **SCREEING TOOLS** that used for accurately screening ASD, including

- ✓ Quantitative Checklist for Autism in Toddlers /Q-CHAT/
- ✓ Modified Checklist for Autism in Toddlers/M-CHAT/
- ✓ Modified Checklist for Autism in Toddlers, Revised, with Follow up/ M-CHAT-R/F /

Con..

- ✓ Screening Tools for Autism in Toddlers and Young Children/STAT/
- ✓ Autism Screening Instrument for Educational Planning,3rd version/ASIEP-3/
- ✓ Autism Behavior Checklist/ABC/
- ✓ Childhood Asperger syndrome Test/CAST/
- ✓ Social Communication Questionnaire/SCQ/
- ✓ Ritvo Autism Asperger Diagnostic Scale Revised/RAADS-R/

Con..

- Diagnose ASD by looking at a person’s behavior and development. ASD can usually be reliably diagnosed by the age of two. It is important for those with concerns to seek out an assessment as soon as possible so that a diagnosis can be made

MINDJOURNAL

Autism Treatment



Behavioral and communication therapy



Educational therapy



Medications

Studies show diet and exercise can help to manage weight, release stress & anxiety and improve social interactions

MIND HELP

Medication

- Use medication to treat some symptoms that are common with ASD. With medication,

A person with ASD may have fewer problems with:

- Irritability
- Aggression
- Repetitive behavior
- Hyperactivity
- Attention problems
- Anxiety and depression

3.7 COMPLICATION

- Sensory problems
- Seizures
- Mental impairment

Other complications that can accompany ASD include

- Diabetes and obesity
- Sleep disorder
- Obsessive compulsive disorder /OCD/
- Social isolation
- Victimization

Thank you



4.Out line

4. Asperger's syndrome

- Definition
- Cause
- Pathophysiology
- Sing and symptoms
- Diagnosis
- Management
- complication


What Is Asperger's Syndrome?

4.1 DEFINITION:

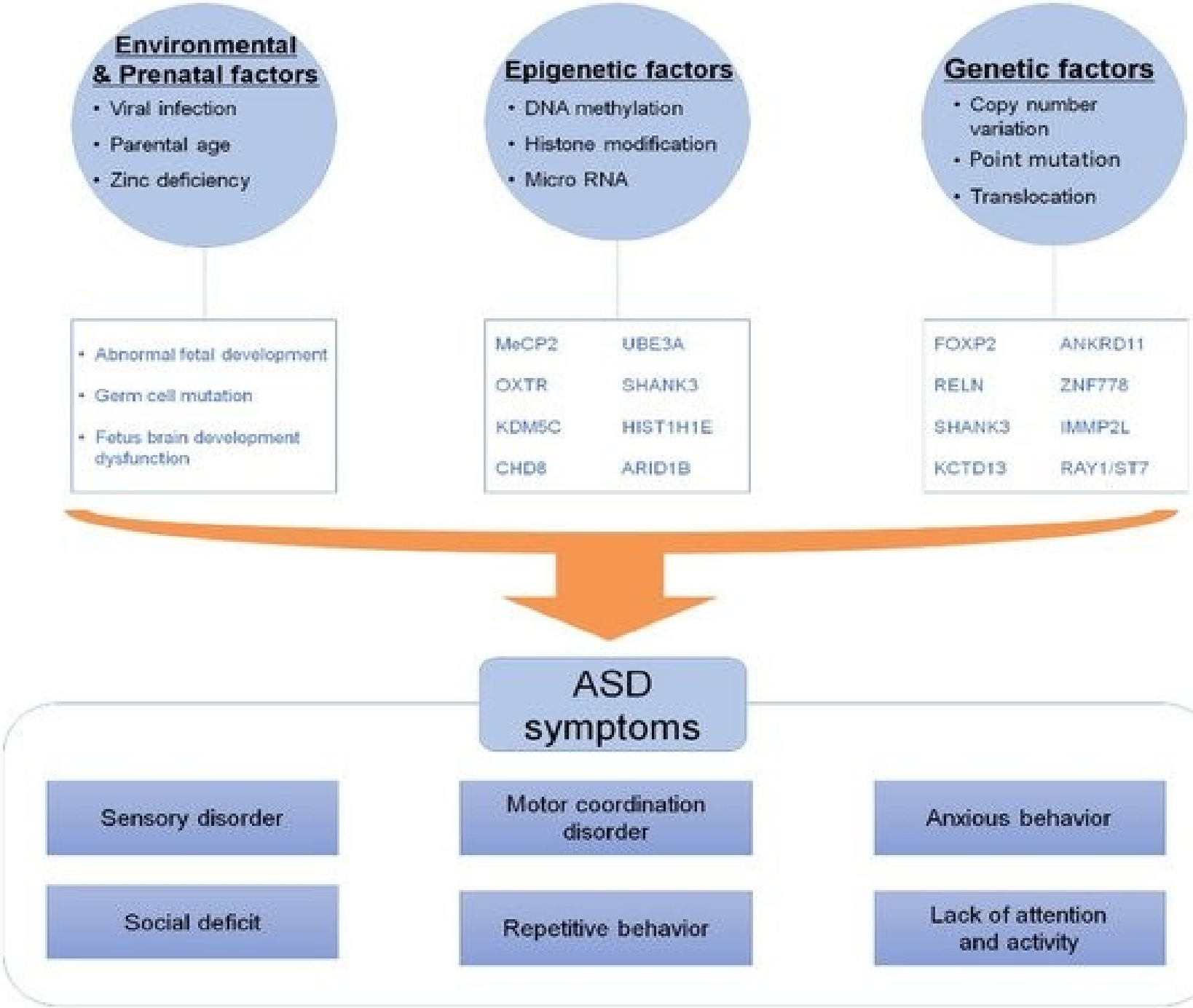
- Asperger syndrome (AS), also known as Asperger’s, is a developmental disorder characterized by significant difficulties in social interaction and nonverbal communication, along with restricted and repetitive patterns of behavior and interests.

4.2 CAUSE

Cause of Asperger syndrome is not yet identified, although an inherited (genetic) component is believed to be involved. In special cases it may caused by pregnancy infections use of teratogens, and exposure to toxics.

- Asperger syndrome may be caused by a combination of different factors like

- Genetic factor
- Environmental factor
- Brain abnormalities


Environmental & Prenatal factors
• Viral infection
• Parental age
• Zinc deficiency
Epigenetic factors
• DNA methylation
• Histone modification
• Micro RNA
Genetic factors
• Copy number variation
• Point mutation
• Translocation
• Abnormal fetal development
• Germ cell mutation
• Fetus brain development dysfunction
MeCP2
UBE3A
OXTR
SHANK3
KDM5C
HIST1H1E
CHD8
ARID1B
FOXP2
ANKRD11
RELN
ZNF778
SHANK3
IMMP2L
KCTD13
RAY1/ST7
ASD symptoms
Sensory disorder
Motor coordination disorder
Anxious behavior
Social deficit
Repetitive behavior
Lack of attention and activity

4.3 SIGN AND SYMPTOMS

- Less social active
- Less friends
- Not interested in making friends
- Inability to express emotions
- No Eye contact
- Less facial expressions
- Inability to use gestures

Con..

- Ineffective communication
- Lack of relation
- Sensitive to external stimuli
- Dependent and
- Repetitive actions like arm waving

MINDJOURNAL

Signs of Aspergers Syndrome



Lack of social awareness



Inability to express thoughts and emotions



Difficulty maintaining relationships



Lack of use and understanding of gestures



Inability to respect interpersonal boundaries



Issues with proximity



Stereotypical & repetitive motor patterns



Lack of or exaggerated facial expressions



Confused & distressed by jokes, sarcasm or irony



Delays in motor skills development



Difficulty with coordination

MIND HELP

Differences between Asperger's and Autism

Asperger's	Autism
Average to above average IQ	Below average IQ
Delayed motor skills	No problems with motor skills
Express interest in others	No interest in others
Normal development of language	Severe lapses in language development

4.4 PATHOPHYSIOLOGY

- The defect is not clearly undrestood.
- Gaigg and Bowler hypothesized that impairments in the connections between the amygdala and associated structures of the brain may play a role in the pathogenesis of Asperger symptoms.
- Dziobek et al. reported elevations of total cholesterol and low-density lipoprotein in people with Asperger syndrome. Dyslipidemia may play a role in Asperger syndrome

4.5 DIAGNOSIS

- diagnosis is the formal identification of the condition, usually by a multi-disciplinary diagnostic team, often including a speech and language therapist, pediatrician, psychiatrist and/or psychologist.
- health history
- psychological and communication assessments

4.6 TREATMENT

- Social skills training
- Speech-language therapy
- Cognitive behavioral therapy (CBT)
- Parent education and training
- Anger management

4.7 COMPLICATION

- Attention deficit hyperactive disorder/ADHD/
- Obsessive compulsive disorder /OCD/
- Anxiety disorder
- Tic disorders, like Tourette syndrome
- Depression
- Disruptive disorder , like aggression, tantrums and self injury

Thank you

