A 4 years old child with 3 main complaints ; Developmental delay , repetitive behaviors and lack of communication with others.

The child does not have proper eye contact with family members or strangers . He does not have sensitivity to sounds and lights . He can not communicate ; he only has blabbering at times of excitement . He has toileting issues and is in the process of toilet training but shows some repetitive characterstics , recognized by parents when he must use the toilet . He uses cell phone and television as per his interests without much assistance . He is interested in music and enjoy listening to some religious songs , old melodies , and rhymes . He also enjoy swimming and playing with puzzles and building blocks . The child is taken out with the family members for family occasions and recreational outing and has been known to adapt with considerable ease . The child prefers to move around on his own but has to be monitored continuously for his safety . The child has been immunized until the date without any side effect . The couple had an arranged marriage and did not have any health issues . The family living in their own house . They have two suns , the child under study is the first child and he has a younger sibling who is 1 years old . The father has been recently diagnosed as a diabetic and the paternal grandmother is diabetic and hypertensive.

There is no history of any other illnesses in the family.

Prenatal and birth history :

mother had a normal pregnancy and attended all antenatal clinic regularly. The child was born by lower segment caesarean (LSCS) after 40 week of gestation when the mother did not have natural labour around the expected date of delivery (EDD).

- At birth , the child cried normally but was pale and shifted to the intensive care unit (ICU) immediately for observation . The child apgar score is not known and his birth weight was 3.2 kgs.
- Breastfeed was not initiated immediately and the child remained in ICU for a few days .
- Growth and development :
- The child growth and development were reported to be delayed.as per reports , there was a slight delay in mental and physical development as compared to normal.
- A few days after birth , it was noticed that the child was flaccid and at three months of age , the child did not have eye contact with the mother .
- The child was examined by the family physician and was reported to have a delay in growth and development and the physical examination showed that the fontanels were closed much earlier .
- They were advised to go for further evaluation . Over the next three years , the child was taken to various hospital , and prominent pediatrician and underwent several therapies and alternative medicine protocols after wich he was assessed and diagnosed as autistic.

- Investigation:

MRI and nerve conduction test were done as part of diagnosis procedure and were returned normal . The child had underwent various therapies like occupational therapy , and alternative medicine protocols including Homeopathy , and acupuncture in healthcare centre and institutes in the local area and places . No medications are given to the child till now . All care for the child is being given by the family and there has been no specific indication of institutionalization for the child .

- Autism spectrum disorder:

Evaluation and diagnosis

استاد راهنما:

آقای دکتر شفیعی – متخصص نورولوژی

عضو هیئت علمی دانشگاه

ارایه دهنده:

دکتر اسمی – دستیار پزشکی خانواده

- **Autism spectrum disorder (ASD) is a biologically based neurodevelopmental disorder characterized by persistent deficits in social communication and social interaction and restricted, repetitive patterns of behavior, interests, and activities.**
- With the ٢٠٢٢ implementation, ICD-١١ changed the terminology for ASD
- ASD is the "parent" term and is further characterized by the presence or absence of a disorder of intellectual development and/or impairment or absence of functional language
- In ICD-١٠, ASD was classified as a "pervasive developmental disorder" with several subtypes (eg, childhood autism, atypical autism, and Asperger syndrome)

- ROLE OF PRIMARY CARE PROVIDER
- Early identification and clinical suspicion
- Refer for comprehensive evaluation
- Make the initial diagnosis
- Refer for intervention pending diagnosis
- COMPREHENSIVE EVALUATION
- Goals
- the child's symptoms meet established diagnostic criteria for ASD
- level of functioning and neurodevelopmental profile
- another condition
- Components – History
- Early ASD symptoms
- ASD symptoms in older children (vary with developmental level at presentation):

- History of common associated conditions:
- Family history
- Psychosocial history
- Examination
- Weight
- Head circumference
- Wood lamp examination
- Examination for dysmorphic features or neurodevelopmental findings
- Examination of muscle tone and reflexes
- Focal neurologic findings
- Diagnostic tools
- Autism Diagnostic Interview-Revised (ADI-R)
- Autism Diagnostic Observation Schedule-2nd edition (ADOS-2)

- Childhood Autism Rating Scale 2nd edition (CARS-2)
- Developmental Dimensional and Diagnostic Interview (3di)
- Diagnostic Interview for Social and Communication Disorder (DISCO)
- Gilliam Autism Rating Scale (GARS)
- Ancillary testing
- Speech, language, and communication assessment
- Developmental/intelligence testing
- Assessment of adaptive skill
- Sensorimotor and/or occupational therapy evaluation
- Vision and hearing assessment
- Lead testing

- DIAGNOSIS
- DSM, Fifth edition criteria
- ICD 11th Revision criteria
- Assessment of severity
- Social communication/interaction(level 1.2.3)
- Repetitive/restricted behavior(level 1.2.3)
- DIFFERENTIAL DIAGNOSIS
- Global delay/intellectual disability - Fetal alcohol syndrome
- Intellectual giftedness - Attachment disorder –Stereotypic movement disorder
- Social (pragmatic) communication disorder - Obsessive-compulsive disorder
- Language disorder - Attention deficit hyperactivity disorder
- Nonverbal learning disorder -Anxiety disorder-Tic disorder/Tourette syndrome
- Hearing impairment - Landau-Kleffner syndrome - Rett syndrome -

- EVALUATION FOR ASSOCIATED CONDITIONS
- Genetic testing
- Initial genetic tests(chromosomal microarray (CMA) and deoxyribonucleic acid (DNA)
- Other testing as indicated
- Metabolic testing
- Neuroimaging
- Electroencephalogram

- SUMMARY AND RECOMMENDATIONS:
- Role of the primary care provider
- Goals of comprehensive evaluation
- Components of evaluation
- Diagnosis
- Additional evaluation

Differential diagnosis of autism spectrum disorder

Condition	Features that may help distinguish the condition from ASD
Global developmental delay/intellectual disability	▪ Social responsiveness and communication appropriate for developmental level
Intellectual giftedness	▪ Normal pragmatic language skills ▪ Intense interests are functional, varied, and can be explained by the child ▪ Social interaction is generally enjoyed
Social (pragmatic) communication disorder	▪ Absence of restricted, repetitive patterns of behavior, interests, or activities
Developmental language disorder	▪ Normal reciprocal social interactions ▪ Normal desire and intent to communicate ▪ Appropriate imaginative play
Language-based learning disorder	▪ Normal reciprocal social interactions ▪ Normal desire and intent to communicate ▪ Appropriate imaginative play ▪ Pragmatic language more typical than in ASD ▪ Desire to communicate (even if competency is lacking)
Nonverbal learning disorder	▪ Impairment in social skills and pragmatic language milder than in ASD ▪ Lack of restricted, repetitive patterns of behavior, interests, or activities
Hearing impairment	▪ Normal reciprocal social interactions ▪ Normal eye-to-eye gaze ▪ Facial expressions indicate intention to communicate
Landau-Kleffner syndrome	▪ Usually have typical development until approximately 3 to 6 years of age ▪ Typically presents with auditory verbal agnosia (behaving as if deaf)
Rett syndrome	▪ Female predominance ▪ Head growth deceleration ▪ Stereotypic hand movements

Fetal alcohol spectrum disorder	▪ Characteristic facial features (not always present): • Short palpebral fissures • Thin vermillion border • Smooth philtrum
Attachment disorder	▪ History of severe neglect or mental health issues in caregiver ▪ Social deficits tend to improve in appropriate caregiving environment
Attention deficit hyperactivity disorder	▪ Normal pragmatic language skills ▪ Normal nonverbal social behavior ▪ Normal imaginative play ▪ Lack of restricted, repetitive patterns of behavior, interests, and activities
Anxiety disorder (includes social anxiety and selective mutism)	▪ Normal nonverbal social behavior and imaginary play ▪ Lack of circumscribed interests ▪ Absence of restricted, repetitive patterns of behavior, interests, or activities
Obsessive compulsive disorder	▪ Normal social skills ▪ Normal pragmatic language ▪ Symptoms are a source of anxiety rather than a pleasure
Stereotypic movement disorder	▪ Normal social skills ▪ Normal pragmatic language
Tic disorder/Tourette syndrome	▪ Normal social skills ▪ Normal pragmatic language

Early symptoms and signs of autism

Caregiver concerns about deficits in social skills
Caregiver concerns about deficits in language skills or behavior
Caregiver concerns about frequent tantrums or intolerance to change
Delayed language and social/communication skills: ▪ Lack of orientation to name by age 12 months ▪ Lack of pointing or gesturing to indicate interest (eg, by pointing to an airplane flying over) by age 14 months ▪ Lack of pretend play (eg, "feeding" a doll) by age 18 months
Avoiding eye contact or wanting to be alone
Having trouble understanding other people's feelings or talking about their own feelings
Repeating words or phrases over and over (echolalia)
Giving unrelated answers to questions
Getting upset by minor changes
Having obsessive interests
Flapping their hands, rocking their body, or spinning in circles
Having unusual reactions to the way things sound, smell, taste, look, or feel

Possible symptoms of autism spectrum disorder in preschool-age children [1-4]

Impairments in social communication and interaction
■ Lack of, delay, or regression in spoken language.
■ If present, spoken language may be atypical, for example: • Unusual intonations (monotone, sing-song) • Echolalia • Incorrect pronoun use (referring to self by name or as "you," "he," or "she" after age 3 years) • Non-speech-like vocalizations (eg, grunting, squealing) • Limited to specific topics of interest
■ Limited use of language for communication (eg, using only single words even though capable of speaking in sentences).
■ Reduced or absent nonverbal communication (gestures, facial expression).
■ Little or no response to others' gestures or facial expression.
■ Lack of response or slow to respond when called by name (despite normal hearing).
■ Reduced or absent interest in or attempts to share interest with another person (eg, by pointing or monitoring the other's gaze), for example: • Lack of social smile or eye contact • Lack of imitation (eg, clapping) • Limited or absent social bids • Lack of ability of interest in sustaining a social interaction • Resistance to being be cuddled
■ Lack of awareness of other people; appearing to be in their own world.
■ Preference for solitary play (lack of initiation or participation in social play with others).
■ Lack of interest in other children or odd social approaches to other children (eg, disruptive, aggressive).
■ Lack of awareness of common social conventions (eg, taking turns in a conversation, awareness of personal space).
■ Lack of or minimal recognition or responsiveness to another's feelings (eg, happiness, distress).

Possible symptoms of autism spectrum disorder in school-age children and adolescents [1-4]

Impairments in social communication and interaction
▪ Abnormal language development, including muteness
▪ If present, spoken language may be atypical; for example: • Unusual prosody of speech (rate, rhythm, tone, volume) • Persistent echolalia • Referring to self by name or as "you," "he," or "she" • Non-speech-like vocalizations • Tendency to speak freely only about specific topics of interest • Talking at others rather than having a back-and-forth conversation • Unusual vocabulary for age or social group • Responses to others may seem rude or inappropriate • Difficulty understanding others' intentions (eg, takes things literally, misunderstands metaphors or sarcasm) • Unable to adapt style of communication to social situations (eg, overly formal or inappropriately familiar) • Reduced and poorly integrated gestures, facial expressions, body orientation, and eye contact
▪ Limited use of language for communication
▪ Reduced, absent, or atypical nonverbal communication (eye contact, gestures, facial expression)
▪ Poor response to name (despite normal hearing)
▪ Little or no response to others' gestures or facial expressions
▪ Reduced interest in people, including children their own age
▪ Apparent preference for aloneness
▪ Difficulty making and maintaining peer friendships (may find it to be easier with younger children or adults)
▪ Reduced or lack of enjoyment of situations that most other children enjoy (eg, birthday parties)
▪ Difficulty joining in play of other children (eg, makes no effort to join in or uses wrong approach [eg, aggressive, disruptive])
▪ Difficulty interacting in unstructured social situations (eg, school recess)
▪ Poor understanding or following of social conventions (eg, greetings, farewell behaviors, taking turns, classroom behavior, awareness of personal space)
▪ Easily overwhelmed by social or other types of stimulation, for example: • Extreme reactions to invasion of personal space • Resistance to being hurried
▪ Reduced or absent response to others' feelings

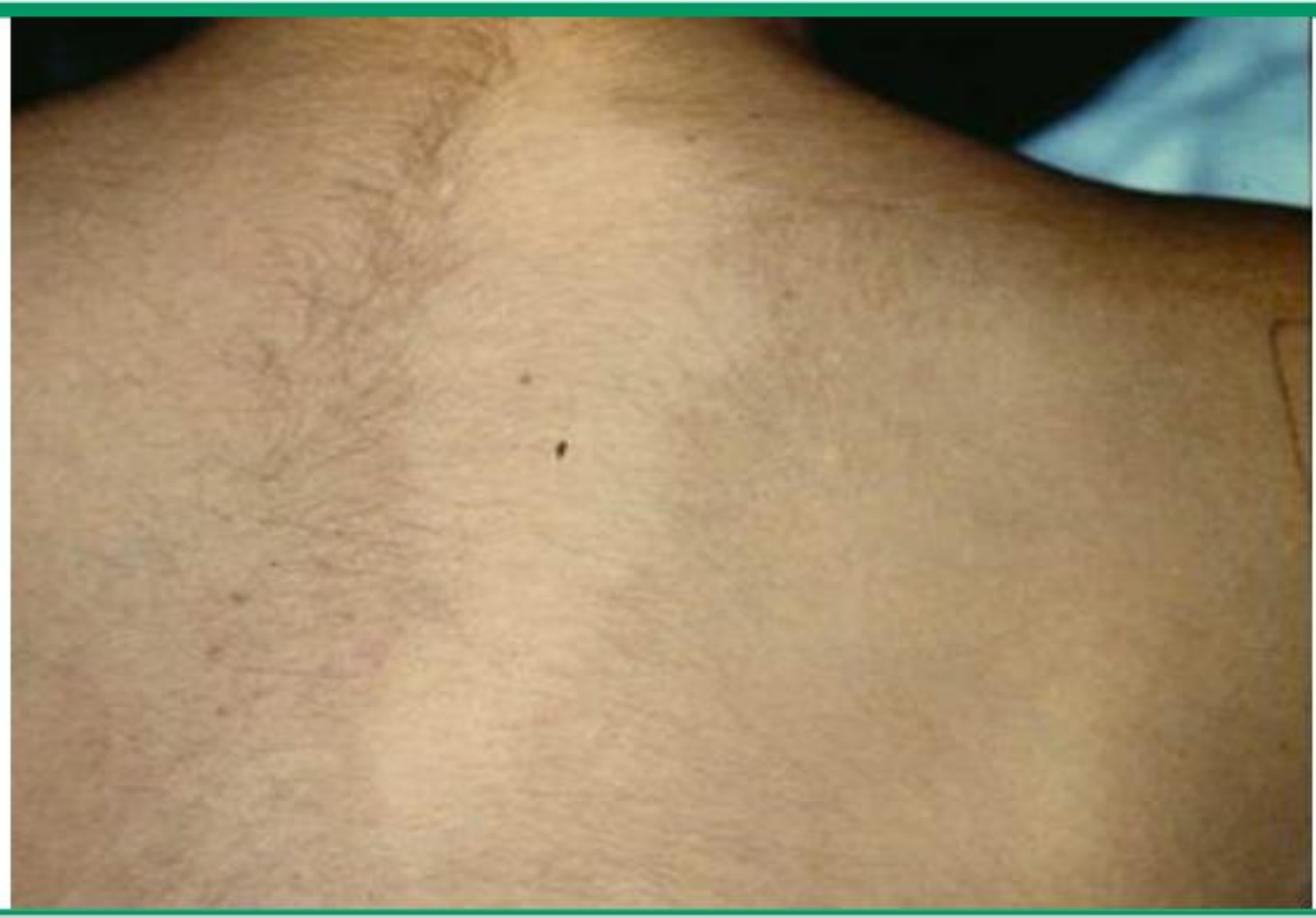
- Extremes of emotional reactivity that are excessive for circumstances
- Abnormal interactions with adults (no interaction or too intense)

Restricted, repetitive behaviors, interests, and activities

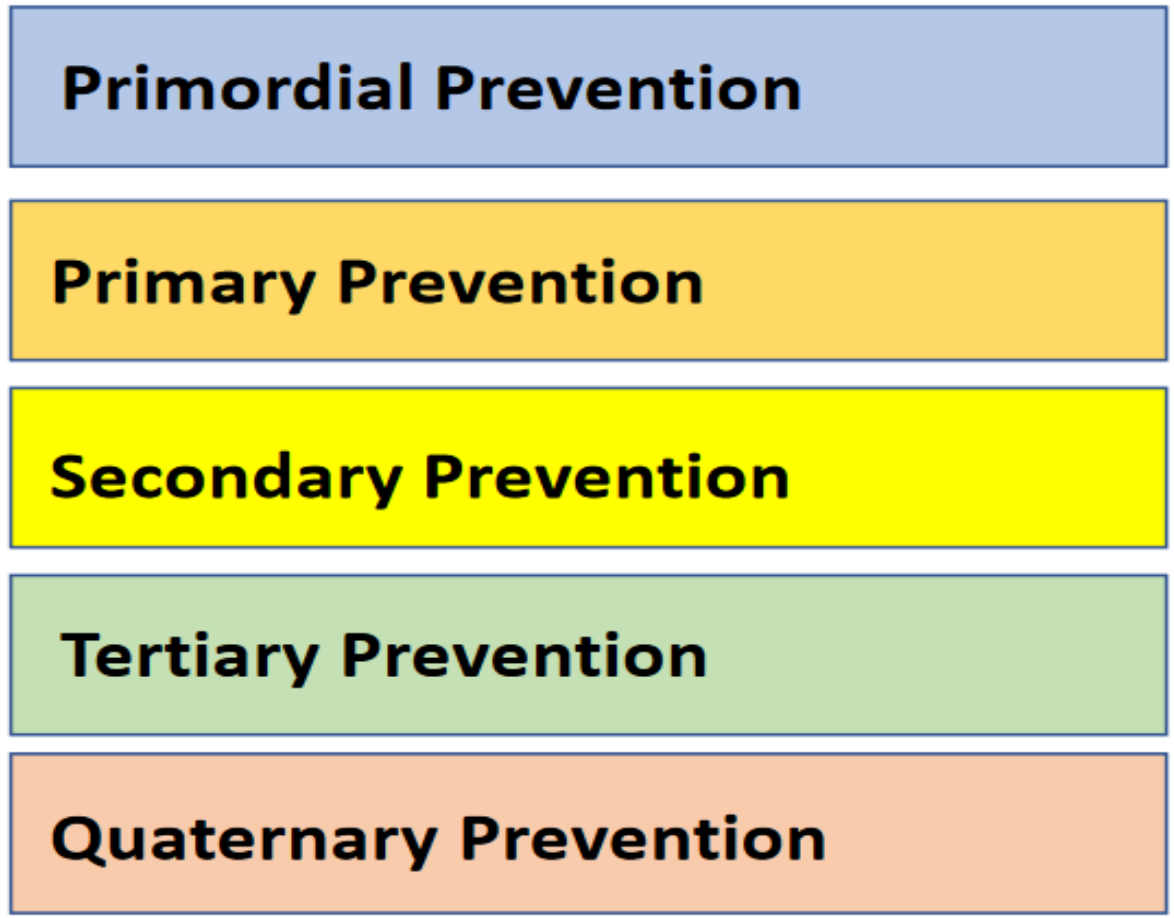
- Lack of flexible, cooperative, imaginative play or creativity, for example:
  - Rigid expectation that other children adhere to rules of play
  - Strong adherence to rules of fairness (may lead to arguments)
  - Repeatedly reenacting scenes from videos or cartoons
  - Preference for highly specific, narrow interests or hobbies (eg, collecting, listing, numbering)
  - Difficulty with imagination (eg, in writing, for future planning)
- Preoccupation with restricted patterns of interest that are abnormal in intensity or focus and interfere with activities of daily life
- Strong preference for familiar routines
- Inability to cope with change or situations that lack structure (may lead to distress [eg, anxiety, aggression])
- Aberrant response to sensory stimuli (over- or under-sensitive), for example:
  - Excessively touching people or objects
  - Preferring to be in the dark

  - Deliberately smelling objects

Tuberous sclerosis complex



Hypomelanotic macule on the torso of a patient with tuberous sclerosis complex.


Primordial Prevention
Primary Prevention
Secondary Prevention
Tertiary Prevention
Quaternary Prevention

Primordial Prevention

- ۱- اقدام در خصوص ترویج سبک زندگی سالم عدم مصرف الکل و سیگار اهمیت ازدواج و فرزند آوری در سن مناسب و اهمیت مراقبتهای سلامتی قبل و حین بارداری
- ۲- آموزش در خصوص تشکیل پرونده الکترونیک سلامت جهت تمامی آحاد جمعیت کشور و ارزش و اهمیت انجام مراقبتهای لازم در هر گروه سنی
- ۳- آموزش های لازم در سطح ملی برای آشنایی با علایم بیماری ریسک فاکتورها

Primary prevention

1. انجام غربالگری و مراقبتهای دوره ای در هر گروه سنی حسب مورد
2. شناسایی افراد پرخطر و در معرض ریسک جهت توصیه های لازم بهداشتی در خصوص سبک زندگی سالم و ترک الکل و درمان بیماریهای همراهی که ریسک ایجاد موارد مثبت را در کودکانشان ایجاد می کند.
3. آموزش سبک زندگی سالم-اجتناب ازآلودگیهای محیطی – سموم – اصلاح اختلالات تغذیه ای در مادر در دوران بارداری – درمان سریع و صحیح بیماریها در دوران بارداری مانند سندرم تورچ –
4. سابقه بیماریهای ژنتیکی در خانواده
5. سابقه تماس با فلزات سنگین مانند سرب
5. سابقه مشکلات حین رایمان
6. سابقه بستری در بخش مراقبت های ویژه
7. سابقه جدایی والدین

Secondary prevention

1.بیماریابی به موقع و انجام غربالگریها قبل از بارداری در جمعیت در معرض ریسک و انجام تستهای بیماریابی و تشخیص اوتیسم

2.غربالگری کوموربیدیتی های زمینه ای

3.درمان سو تغذیه مادر باردار

4.درمان به موقع بیماریهای دوران بارداری مانند تورچ

5.انجام مراقبت های کودک اوتیسم در مراکز بهداشتی و معرفی به کلینیک های بیلی و ویزیت متخصص اطفال و نورولوژی

Tertiary Prevention

- ۱- درمان بموقع و مقتضی براساس آخرین و جدیدترین مطالعات
- ۲- درمان کوموربیدیتی های همراه واقدامات پیشگیرانه جهت کنترل بیماری
- ۳-مراقبت و مونیتورینگ بموقع بیماران

Quaternary Prevention

- ۱- مونیتورینگ و فالواپ بموقع بیماران و ارایه خدمات درمانی مقتضی
- ۲- عدم انجام اقدامات پاراکلینیکی و دارویی که تاثیر خاصی بر پیش آگهی و عوارض بیماری ندارد