

## Introduction The consequences of disease

 Something abnormal Etiology (At birth or acquired)

Changes in structure Path
 Or function ↓

↓ Pathology ↓

Manifestation

Symptoms & Signs

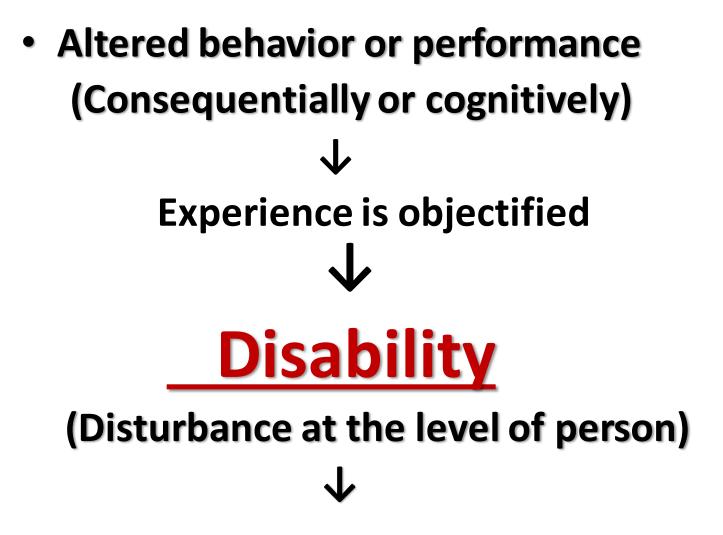
## Someone becomes aware Clinical disease

#### Pathological state is exteriorized

## $\checkmark$

## **Impairment**

#### (Disturbance at the organ level)



# Disadvantage relative to others Socializing the experience ↓



## Definitions

## Impairment:

- In the context of health experience, Impairment is any Loss or Abnormality of physiological, psychological or anatomical Structure or Function
- It refers to an underlying molecular, cellular, psychological or structural disorders within an individual.

## Definitions

## Disability

- In the context of health experience, Disability is any <u>Restriction or Lack</u> of <u>Ability to Perform an Activity</u> in the <u>Manner</u> or within the <u>Range</u> considered normal for a human being.
- <u>It refers to a Stable and Persistent deficit in</u> <u>function</u>, often the consequence of Impairment and also confined to the individual.

## **Definitions** Handicap:

- In the context of health experience, A handicap is a <u>disadvantage for a given individual</u>, resulting from <u>an impairment or a disability</u>, that <u>limits</u>
   <u>or prevents the fulfillment of a role that is</u> <u>normal for that individual (depending on age,</u> sex, and social and cultural factors).
- It refers to the limitation on carrying out social roles defined as appropriate for an individual of certain age, gender or class.

## Definitions

## New definition and classification:

- The new definition emphasized that, <u>the human</u> <u>functioning state</u> is a result of the interaction between three dimensions:
- 1. The biological status with
- 2. the psychological status and
- 3. the coping mechanisms of the human being and the environment and the society surrounding.

#### New definition and classification:

- In the past, disability was interpreted according to a <u>Medical Model</u>. That is, disability was linked to various medical conditions, and was viewed as a problem residing solely (exclusively) in the affected individual.
- Disability was seen solely as the result of an individual's inability to function.

#### New definition and classification:

- ✓ This medical model has recently been replaced by the Social Model of disability.
- Social model conceptualizes disability as arising from <u>the</u> interaction of a person's functional status with the physical, cultural, and policy environments.
- ✓ If the environment is designed for the full range of human functioning and incorporates appropriate accommodations and supports, then; People with functional limitations would not be "disabled" in the sense that they would be able to fully participate in society.









## **Required Interventions**

- Three major areas of required interventions; namely,
- 1. Prevention
- 2. Rehabilitation
- **3. Equalization of opportunities**

## **Required interventions**

## **A-Prevention:**

 All the measures aimed at <u>preventing the</u> <u>onset</u> of mental, physical and sensory impairments (<u>primary prevention</u>) or

 Measures aimed at preventing impairment, when it has occurred, from having negative physical, psychological and social consequence.

## **Required interventions**

#### **B-Rehabilitation:**

- Aimed at enabling an impaired person to reach an optimum mental, physical and social functional level, thus providing him with the tools to change his own life.
- It can involve measures:
- intended to compensate for a loss of function and/ or intended to facilitate social adjustment or readjustment.

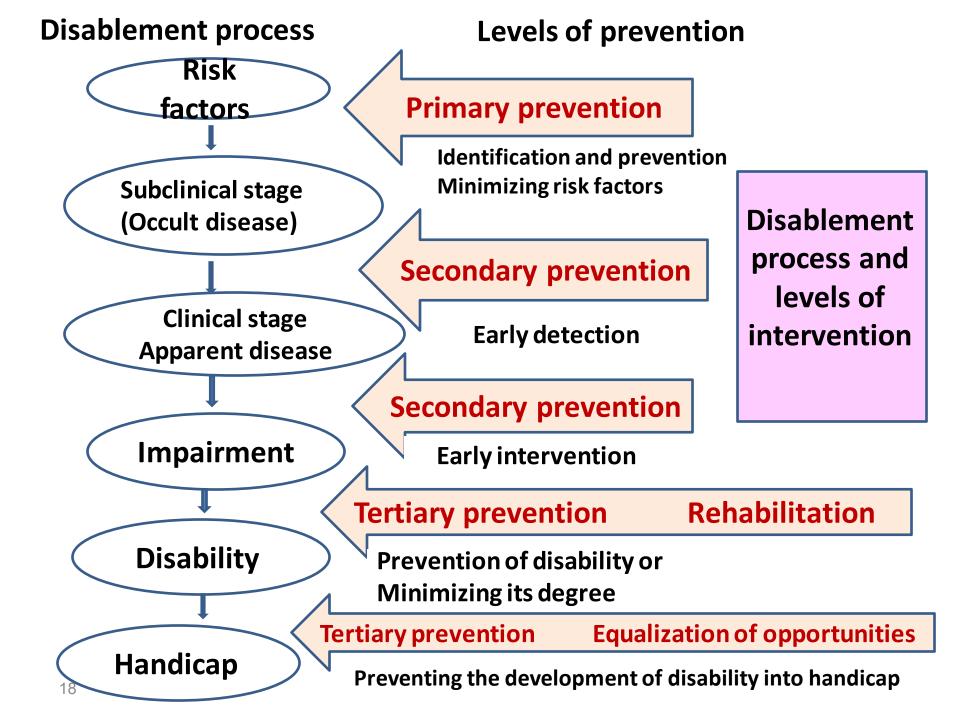
## **Required interventions**

#### **C-Equalization of opportunities:**

It is the process through which the

general system of society are made accessible to all such as:

- 1. The physical and cultural environment,
- 2. Housing and transportation,
- 3. Social aid health services,
- 4. Educational and work opportunities,
- 5. Cultural and social life, including sports and recreational facilities, are made accessible to all.



## **Benefits of early intervention**

Helps children with disabilities: Gains in cognitive, physical, language and social skills.

#### **Benefits for families:**

- ➢ Helps families manage the child
- ➢ Reduces stress.

#### **Benefits for the society:**

- ➢ Reduces means for institutional placement.
- ➢ Reduces need for special education

#### Saves money

#### I. Emotional adjustment

People's reaction to their disabilities vary with:

- A- <u>The individual</u>, the <u>severity</u> of the handicap, and the <u>time</u> of life when the condition began
- **B-** <u>Social</u>: social discrimination and the limited <u>concessions</u> made by the community

Individuals born with an impairment:

 Those people have <u>never experience</u> what we consider a normal life, and their awareness that they are <u>different</u> may develop slowly as their social contacts increase.

#### Individual who acquire an impairment:

(through injury or sudden disease have more dramatic and immediate adjustment to make)

- They must cope with their medical problems.
- They have to adjust to a new social situation and accept new attitudes from others.
- In addition they may be coping with job loss, family crisis, and marital stress.

#### II. Social issues:

1. <u>The attitude of others:</u>

#### This is a major problem due to:

- A. lack of understanding on the part of the non-disabled and , on the other hand
- B. there is limited patience on the part of the handicapped individuals.

Disabled may react with <u>anger and frustration</u> when <u>inappropriate assistance</u> is forced upon them from non-disabled persons.

- 2. <u>Behavioral and social acceptance:</u>
- Social behavior skills (مهارات السلوك الاجتماعي) are needed for a person to experience social acceptance and participation in community life.
- This is considered to be a difficult task in disabled persons.
- In order to have a socially productive life disabled persons face three main problems:
- 1. Affordability and availability of assistive devices.
- 2. Employment and financial concerns.
- 3. Accessibility problems, e.g. transportation.

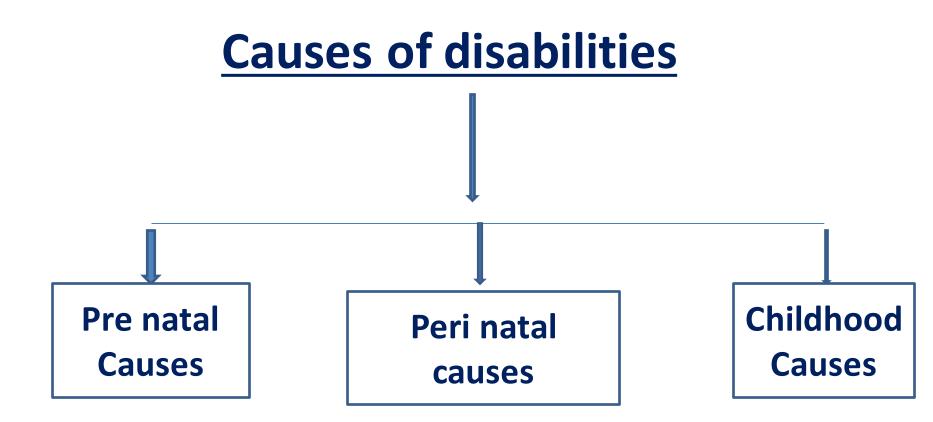
## **Categories of childhood disabilities**

## A) Sensory disabilities:

- 1. Visual disabilities (partial sight or with blindness)
- 2. Hearing disabilities (lack or reduction in the ability to hear clearly)
- 3. Speech or language impairment (which has an impact and an adverse effect on the child educational performance).

## **Categories of childhood disabilities**

- **B)** Physical (motor) disabilities
- C) Mental disabilities: as mental retardation
- **D)** Emotional disturbance
- E) Specific learning disability
- F) Traumatic brain injury
- G) Other health impaired conditions, e.g. chronic diseases as DM, epilepsy.



#### 1) <u>Chromosomal cause:</u>

 Chromosomal abnormality can involve the <u>loss, gain or</u> <u>exchange of genetic material</u> from chromosome pair, e.g. <u>Down syndrome</u>.

#### 2) Genetic causes:

• e.g. genetic defect as <u>sickle cell disease</u>.

#### 3) <u>Rh. Factor:</u>

 When an Rh +ve man and an Rh -ve woman have children with <u>Rh incompatibility</u>, the mother's blood may begin to form antibodies against the foreign positive Rh factor.

# The resulting destruction of Rh incompatibility may be:

- <u>Limited</u> causing only anemia,
- <u>Excessive</u> causing cerebral palsy, deafness, mental retardation or even death.

## 4) Maternal stress:

Prolonged emotional stress during pregnancy may have a consequence on the child <u>as low birth weight (LBW)</u>, <u>hyperactivity and irritability</u>.

#### 5) Environmental causes:

a) **External agents:** as exposure to irradiation such as

X ray, the greater danger for malformation comes between the <u>second and sixth weeks</u> after conception.

#### b) Drugs: Teratogens are drugs producing birth defects as

- 1. Legal drugs (nicotine, caffeine),
- 2. Prescription drugs (some antibiotics, hormones, steroid, and tranquilizers)
- 3. Illegal drugs (Cocaine, heroin, marijuana and environmental pollutants (lead, methyl mercury)

- c) <u>Maternal nutrition</u>: Example deficiency of <u>Folic Acid</u> is a risk factor for having a baby with <u>Spina Bifida.</u>
- d) <u>Maternal infections:</u> viral diseases such as cytomegalovirus, rubella and chicken pox are particularly dangerous during the early fetal periods.

- 6) Maternal diseases and disorders during pregnancy:
- Children whose mothers had <u>severe toxemia</u> during pregnancy have a risk of lowered intelligence.

#### 7) Age of the mother:

- <u>Teenage</u> mothers have a greater risk to have babies with <u>low birth weight</u> and <u>neurological defects</u>.
- Mothers <u>over 40</u> have an increased risk of having children with <u>chromosomal abnormality</u>.

- **1. Drugs** taken during labor and delivery as some <u>painkillers</u>.
- 2. Premature infants: this is more common among:
- economically deprived mothers
- smoking
- alcohol
- various drugs

## Peri natal causes of disabilities

## 3. Oxygen deprivation:

- brain hemorrhage
- failure to breath
- more likely to occur with prolonged labor
- Infections: sexually transmitted diseases
   (STD) can affect the baby (example; AIDS, Gonorrhea, Syphilis and herpes)

## III. Childhood causes of disabilities

- 1) Injuries
- 2) Childhood diseases: Meningitis and encephalitis
- 3) Environmental causes: for some children environmental deprivation has a debilitating effect on the development of abilities such as Language Use, Adaptive Behavior, and Cognition
- Deprivation can include: poor nutrition, poor housing, lack of social interaction







#### **Primary prevention:**

- **A)** Genetic counseling: It is the process of providing information on:
  - **1.** The nature and consequences of genetic disorders i.e. it has a diagnostic aspect
  - **2. Genetic recurrence risk** i.e. to ascertain which individuals at risk of having an affected child
  - 3. The means available for the prevention of transmission of defective genes

 <u>Genetic screening and counseling prior to</u> conception is important for the control of <u>genetically determined disabilities</u>

#### B) Pre-pregnancy Planning: as checking

medical conditions of women, Rh factor, infections such as <u>venereal diseases</u> and rubella immunization

c) Improved prenatal, natal and postnatal care.

#### The aspect of prevention concentrates on:

- ✓ The management of maternal risk factors at the time of delivery
- ✓ Support for the premature or compromised neonates

D) Immunization programs: Programs of general immunization during infancy have led to a remarkable decrease in infectious diseases that used to be a major cause of disability

• These include Poliomyelitis, Tuberculosis, Meningitis, Encephalitis and Measles.

#### **E)** Regulations & legislation

# Certain health regulations and legislation such as:

#### ✓ Mandating immunization of infants

 Legislation concerning safety of toys, seatbelts, fireworks...etc. also play an important role in primary health prevention

#### **Secondary prevention**

- Accurate and early recognition of the disability:
- Some impairment features are physical & obvious during clinical examination
- These include skeletal abnormalities, blindness, hearing and speech disorders, some mental disorders and some <u>chromosomal anomalies</u>

#### FIGURE 2.4 - SOME CHROMOSOME ABNORMALITIES

Name	Description	Treatment	Incidence
Down syndrome	An extra chromosome causes mild to severe retardation and physical abnormalities.	Surgery, early intervention, infant stimulation, and special learning programs	1 in 1,900 births at age 20 1 in 300 births at age 35 1 in 30 births at age 45
Klinefelter syndrome (XXY)	An extra X chromosome causes physical abnormalities.	Hormone therapy can be effective	1 in 600 male births
Fragile X syndrome	An abnormality in the X chromosome can cause mental retardation, learning disabilities, or short attention span.	Special education, speech and language therapy	More common in males than in females
Turner syndrome (XO)	A missing X chromosome in females can cause mental retardation and sexual underdevelopment.	Hormone therapy in childhood and puberty	1 in 2,500 female births
XYY syndrome	An extra Y chromosome can cause above-average height.	No special treatment required	1 in 1,000 male births

✓ Several other disorders do not become evident until later in life.

 This has been possible using biochemical tests and more recently, by applying recombinant DNA technology to the identification of the molecular basis of genetic disorders

#### **Responsibility of early detection**

- The family in general and the mother in particular play a significant role in early detection of disability
- Abnormalities in development, both physical and others and delay in learning abilities may become obvious to the diligent eyes of the mother much earlier than a clinical examination can made

- Besides the parents and the medical team in the hospitals, or the child health centers, day care nursery staff,
- Teachers at nursery schools and school teachers, school nurse, school medical officer, social workers, educational psychologist...etc. are involved in <u>early diagnoses</u>

Strategies on the secondary prevention level can be applied either at the:

- Prenatal or neonatal level
- Some of the conditions that can be diagnosed during the prenatal and neonatal stages are listed in table 1

## Table 1. Tests used in early identification of disabilities or diseases that may lead to disabilities

Stage	Test	Diagnosis
	Maternal &-fetoprotein	
	- Elevation	Neural tube defect
	- Decrease	Trisomy 21, trisomy 18 (Edwards syndrome)
Prenatal	Maternal un-conjugated estral decrease	Trisomy 21, trisomy 18
	Maternal human chorionic gonadotropine elevation	Down syndrome
	Ultra-sonography	Down syndrome and other physical defects
	DNA analysis of amniotic fluid, chorionic villi or fetal blood	Several inborn errors of metabolism
	Cytogenetic	Numeric and structural anomalies of the chromosomes

Table 1. Tests used in early identification of disabilities or diseases that may lead to disabilities			
Stage	Test	Diagnosis	
Neonatal	Estimation of phenylalanine	Phenyl-ketonuria	
	Estimation of other amino-acids	Amino aciduria	
	Estimation of thyroid hormone level	Hypothyroidism	
	DNA analysis	<ul> <li>Several inborn errors of metabolism</li> <li>Cystic fibrosis</li> <li>Thalassaemia</li> <li>Other haemoglobinopathies</li> <li>Duchenne muscular dystrophy</li> <li>Hemophilia</li> </ul>	

- Neonatal screening : <u>organized</u> examination of <u>all neonates</u> in order <u>to diagnose</u> specific disorders so that they <u>can be treated</u>
- It is an established preventive approach and includes both clinical and biochemical screening and have been successful in reducing disability
- Examples of the usefulness of such action are shown in table 2

#### Table 2. Usefulness of early detection and intervention programs

Disorder	Intervention	Benefit
Hypothyroidism Phenyl-ketonuria	Hormone replacement therapy Low phenylalanine diet	Prevention of mental retardation Prevention of mental retardation
Congenital heart diseases	Surgical correction	Normal growth and development
Familial growth hormone deficiency	Hormone replacement therapy	Normal growth and development
Familial Hypercholesterole mia	Lowering plasma low density lipoprotein level by drugs and diet restriction	Prevention of heart diseases
Hemophilia A	Factor VIII replacement	Avoidance of hemorrhage

Disorder	Intervention	Benefit
Infectious diseases:		
-Polio myelitis	Vaccination	Avoidance of skeletal disability
-Meningitis	Vaccination	Avoidance of mental disability
-Sickle-cell disease	Proper care and management	Normal growth and decrease crisis
Thalassthaemia major	Blood transfusion and iron chelation therapy	Normal growth and development
Congenital dislocation of the hip	Surgical or physical correction	Normal movement
Cleft lip and cleft palate	Surgical correction	Normal facial appearance

- New forms of secondary prevention, such as genetic or surgical manipulation of an affected fetus to eradicate the biochemical or anatomical abnormality are being tried.
- This is true for congenital heart disease (VSD, ASD), cleft lip and cleft palate, congenital dislocation of the hip (CDH) and others

#### **Tertiary Prevention (Rehabilitation)**

- Rehabilitation is the process aiming at enabling an impaired person to reach an optimal (mental, physical and / or social) functional level, thus providing him with the tools to change his own life
- It includes all the services provided to enable the handicapped to make <u>maximum use of</u> <u>their abilities</u>







#### **Tertiary prevention (rehabilitation)**

#### It includes the following measures:

- Measures to compensate for the limitation or loss of function (e.g. by technical aid)
- 2. Measures to facilitate social adjustment or readjustment





 Through these measures the highest possible degree of "independence" can be achieved which usually includes engagement in suitable work

#### **Tertiary prevention (rehabilitation) Special success of rehabilitation depends on:**

- ≻Age
- >Type of home
- Belief implicitly in the success (that further progress is possible)





Personal factors which motivate the patient and family towards overcoming the effects of disability (self-pity-overprotection)