

CEREBRAL PALSY

Cerebral palsy is defined as non-progressive neuromotor disorder of cerebral origin due to abnormal brain development or brain damage in fetal life, during delivery, at birth and early infancy.

Causes

- Premature babies
- Lack of oxygen to the brain at birth
- Severe jaundice during first week of life
- Metabolic disorders (low blood glucose)
- Infection of the brain
- Intracranial hemorrhage.

Signs & Symptoms

- Convulsions
- Visual & hearing defects

- Mental subnormality
- There is a stiffness or spasticity of limbs (more in lower limbs which may cross each other like a scissor) with inability to sit, stand, walk and difficulty in holding objects
- The speech is often delayed or lack clarity
- Irregular movements or tremors of the limbs.

Early Markers of Cerebral Palsy

It is possible to make an early diagnosis of cerebral palsy by 4 months of age by looking for following clinical markers.

- Episodes of inconsolable and unexplained crying, feeding difficulties, lip smacking or chewing movements, excessive sensitivity to light & sound producing startle response
- Persistent neck tonic or

- asymmetric posture beyond 4 weeks of age
- Absence of social or interactive smile by 6 weeks
 - Clenched fists with thumbs adducted & flexed across the palms (cortical thumbs) beyond 8 weeks.
 - Paucity or absence of cycling or fidgety limb movements during first 6-12 weeks of life
 - Lack of stable head control or rolling over by 4 months.
 - Slow head growth

Management

A great deal can be done with the help of appropriate physiotherapy by ensuring proper movements and exercises of different groups of muscles. Massage should not be done as it may further increase muscle tone and spasticity. The child should be

Date: / /
Page: 25

made self-reliant to feed, bath & dress himself even when he takes a long time & does it awkwardly. Some children with cerebral palsy have unique or specialized capabilities like interest in painting & instrumental music. Parents & health care professionals must identify these & other special attributes & harness them effectively by guiding & inspiring the child. After getting hands-on instructions from a physiotherapist, family should continue the exercises & manipulations at home.

Drugs & surgical procedures are available to reduce spasticity. These children must be provided with routine immunizations as per recommended schedule. Due to swallowing difficulties, they may have to feed with a liquid or semi-liquid homogenized diet. Prokinetics (domperidone)

Date: / /

Page:

& ranitidine are given for treatment of feeding difficulties. Avoid overfeeding because they have a tendency to become obese due to lack of physical activity.

CHROMOSOMAL ABERRATIONS

- Chromosomal aberrations are disruptions in the normal chromosomal content of a cell

& are a major cause of genetic conditions in humans, such as Down syndrome.

- In other words, they are changes in the number and/or arrangement of genes in the chromosomes.
- Change in number of chromosomes is known as aneuploidy or numerical aberration.
- Change in arrangement of genes in the chromosomes is known as structural aberration.
- Chromosomal aberrations may involve changes in single chromosome, known as intrachromosomal aberrations.
- Chromosomal aberrations may involve changes in two chromosomes, known as interchromosomal aberration.

Each chromosome of a species usually remains same generation after generation. Variation

in the structure of chromosome which can be produced artificially by radiations, chemicals. Temperature is known as artificial chromosomal aberrations.

The mechanism of structural aberration consists of the breaking & rejoining of chromosomes resulting in deficiencies, duplications, inversions and translocation.

I. Change in Chromosomal Number (Aneuploidy) [Numerical aberration]

- * Monosomy: One chromosome is missing

e.g. Turner's syndrome

- * Trisomy: One extra chromosome is present.

e.g. Down syndrome.

- * Tetrasomy: Two extra chromosomes are present.

e.g. Cell contain 48 chromosomes.

- * Triploidy: One extra chromosome set of haploid genome is present.

- e.g. Cell contain ~~60~~ 69 chromosomes
- * Tetraploidy: Two extra chromosome sets of haploid genome is present.
e.g. Cell contain 92 chromosomes

II. Major changes in chromosome structure (structural aberrations)

→ Deficiencies or Deletion

A
B
C
D
E
F
G
H
I
J

A
B
C
E
F
H
I
J

Loss of the segment of a chromosome is called deletion or deficiency. They are of two types:

- Terminal
- Interstitial/intercalary

→ Duplications

Segment of a chromosome is repeated in a duplication. There are three types of duplications:

- Tandem
- Reverse tandem
- Displaced

a) Tandem

When the duplicated segment

A	B	C	D	E	F	G	H	I	J
A	B	C	D	E	F	E	F	G	

adjacent to its homologous region in the same orientation.

b) Reverse tandem

When the duplicated region is adjacent to its homologous region but in the reverse orientation.

c) Displaced tandem

When the duplicated segment is at different region or position of the chromosome either in the same arm or in other arm.

→ Inversion

When the segment of the chromosome rejoin at the same place but in the reverse orientation.

It consists of two types -

a) Paracentric inversion

b) Pericentric inversion

a) Paracentric

When the centromere is not included in the inversion.

b) Pericentric

When the centromere is included in the inversion.

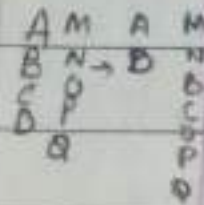
=> Translocation

When a broken segment of a chromosome gets integrated into a non-homologous chromosome:

It is of two types:

a) Simple translocation

When segment of one chromosome integrates into another non-homologous chromosome.



b) Reciprocal translocation

When there is no change in the total amount of genetic material but two non-homologous chromosomes exchange their segment (genetic material).

