

SESI 7

PERTUMBUHAN ANAK, MASALAH pada BAYI dan KANAK-KANAK

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DESKRIPSI

Materi membahas tentang development mile-stones patokan pertumbuhan dan perkembangan bayi dan anak, ASI dan kemajuan perkembangan pertumbuhan bayi dan kanak-kanak, faktor-faktor yang berpengaruh, dan berbagai kondisi gangguan umum kesehatan bayi dan masa kanak-kanak, kondisi gangguan kromosomal, Down's Syndrome & Autism

TUJUAN INSTRUKSIONAL UMUM

Memahami:

- peringkat pengembangan kemampuan dan pertumbuhan bayi dan kanak-kanak serta faktor-faktor yang mempengaruhi - pengembangan keterampilan bayi dan kanak-kanak
- neonatal dan pentingnya ASI bagi kesehatan anak
- berbagai jenis gangguan pada bayi dan kanak-kanak.
- gangguan kromosomal, Down's Syndrome & Autism
- *Genetic Disorders*

TUJUAN INSTRUKSIONAL KHUSUS & POKOK/SUBPOKOK BAHASAN

Menjelaskan tentang:

- Development Miles-stones dan faktor-faktor yang mempengaruhi
- Cara pengembangan keterampilan bayi dan kanak-kanak
- Neonatal , pemberian ASI dan pertumbuhan bayi
- Gangguan pada bayi dan kanak-kanak.
- Patologi Down's Syndrome dan Autism

READING 1 DEVELOPMENT MILE-STONES

Children acquire physical, mental and social skill in well-recognized stages called:

“ development mile-stone”.

*Although there is wide variation in the rate at which each child progresses, **most children develop certain skills by a predictable age.***

FACTORS AFFECTING DEVELOPMENT

*A child becomes capable of developing certain skills only as his or her **nervous system matures.***

*The rate at which maturity occurs is determined generally for each individual and **modified by environmental factors in the uterus and after birth.***

*For instance girls often begin to walk and/or talk at an **earlier stage than boys.***

FACTORS AFFECTING DEVELOPMENT (Cont.-1)

*Premature children miss out on some growing time in the uterus; the time they take no progress should be calculated from the **full-term pregnancy date**, not the actual date of birth.*

*Sight and hearing are both crucial to a child's general development progress, any **defect will affect the child's ability to watch, listen, learn and imitate.***

FACTORS AFFECTING DEVELOPMENT (Cont.-2)

***Intelligence** also affects a child's development, especially in the:*

- ***acquisition of speech and***
- ***ability to coordinate muscles** for precise movements, such as holding a pencil.*

***The home environment** plays an important part in developing the child's potential for certain skills.*

FACTORS AFFECTING DEVELOPMENT (Cont.-3)

Speaking to and playing with children is essential for:

- ***language development and***
- ***practicing new physical skills.***

*Introducing children to other children at the age of 2 or 3 years provides them with **plenty of stimulation.***

HOW A CHILD DEVELOPS SKILL

*Reflex actions present at birth **gradually disappear** as the child learn to **perform voluntary actions** and develops sufficient muscle strength and control to perform them.*

*Often, the child's action progress from seemingly unconnected movements to the **ability to control** part of his or her body.*

*In most children development begin with **control of the head and progresses down the body** untill control of the arms, trunk, and legs is attained.*

HOW A CHILD DEVELOPS SKILL (Cont.-1)

Walking is achieved in numerous stages:

- *from lying with head raised,*
 - *to sitting unsupported,*
 - *to crawling,*
 - *to toddling,*
 - *to standing,*
- and usually*
- *to walking unaided.*

HOW A CHILD DEVELOPS SKILL (Cont.-2)

*A baby begins to develop **hand-eye coordination** from birth.*

*He or she watch objects, learns to focus and to judge distance, and develops the connection of seeing and doing by **watching his or her hands**.*

*Both hand-eye and body limb coordination can be encouraged in an older child (by **practicing ball games**, for example).*

HOW A CHILD DEVELOPS SKILL (Cont.-3)

*At birth, a child communicates his or her needs by **crying**.*

*After vision and hearing are sufficiently well developed, a child watches the parent's mouth intently to learn how to **smile and listens** to the parent speaking before attempting to imitate sounds.*

*A child is able to **concentrate on learning only one skill at a time**, often forgetting a recently mastered skill that will appear again some time in the future.*

DEVELOPMENTAL MILESTONES

- *When assessing the development of a child, specialists in child development look at abilities in 4 (four) main areas:*
 - (1) *locomotion*
 - (2) *hearing and speech*
 - (3) *vision and fine movement, and*
 - (4) *social behavior and play.*

DEVELOPMENTAL MILESTONES (Cont.)

All children acquire skills in much the same order – for example:

A child will not stand before learning to sit.

*The rate at which these skills are acquired varies enormously; a more detailed professional investigation is necessary only if a **child's progress is significantly out of line or if the parent is concerned for some other reason.***

(Sumber AMA Medical Encyclopedia)

READING 2. ***THE NEW BABY***

- *Despite being dependent on its parents, a new baby has some **in-build abilities** that help it along as it develops.*
- *When a new baby enters the world, it is for the first time a separate human being, although it is still entirely reliant on its parents **for warmth, protection, and food.***

*From the time of birth, the developing baby makes rapid advance **toward becoming truly independent.***

READING 2 (Cont.-1)

- *By the age of 18 months or so most children can **walk, talk, feed themselves, and handle a whole range of practical everyday problems.***

*The newborn baby can see and hear the world around it and has an **efficient sense of touch** .*

*But muscular coordination and strength are **not well developed** to begin with.*

READING 2 (Cont.-2)

*It does, however, have some **reflex movements that are valuable for survival.***

*For instance, a baby **cries when tired, uncomfortable, or hungry**, and parents find crying almost impossible to ignore.*

*The **suckling reflex is also vital** – when a baby cheek or lips are touched, it turns at once toward the touch, grasps the object in the lips, and begin sucking.*

*This specific behavior pattern allows efficient feeding from the **nipple of the mother or a bottle.***

READING 2 (Cont.-3)

- *More sophisticated movement (motor) skills, culminating in **self-feeding and unaided walking**, develop as the baby's muscles and nervous system mature.*

At birth, for instance, the nerves that control muscle contraction are not insulated by the usual fatty sheath of myelin. But by 12 month, almost all nerves areased in myelin, aiding walking and other feats of motor control.

Repetitions and practice help solve coordination problems.

There is no fixed timetable

- *Or even strict order in which a developing baby is able to perform particular activities.*

The exception is walking, in which stages are attained in a relatively set pattern and at fairly predictable times.

*After birth, a baby is unable to lift its head, but **4 to 6 weeks later**, increasing muscle strength means it can raise its head from the horizontal.*

- ***At 4 to 5 months.****a baby can lie on its front and lift its bottom up by pushing its knees forward or lift its shoulder from the ground by pushing up with the arms*

READING 2 (Cont.-4)

- ***By 9 or 10 months**, many babies can crawl on knees and hands. Others use “bottom shuffling” derived from sitting skills.*
- ***At around 11 months**, many children can pull themselves into standing position, either with help from an adult or by hanging onto furniture or other supports.*

But staying upright is difficult, and babies at this age tend to wobble and fall over if they attempts to move.

READING 2 (Cont.-5)

- *Once a child is accustomed to the idea of taking all its weight on its feet, its confidence soon grows, and **standing, the preliminary to walking, becomes second nature.***
- *However, at this stage a child might have **trouble sitting down again.** A child set down on the floor or in a crib will pull up to the standing position and then get stuck and cry for help.*

This stage only lasts for a few weeks, and with help – a gentle as lowering onto the floor – the child soon learns how to sit down again without a hard bump.

READING 2 (Cont.-6)

- *By about a year, many babies are able to walk in a coordinated fashion by placing one leg in front of the other in a patterned way. Normally, though, they will need a mobile support, such as an adult's hand or a walker, to do this without falling. The first unaided step soon develops into a sequence of steps, and the child becomes mobile.*

Some children seem to skip stages when learning to walk; it is not uncommon for a child to go from the precrawling stage straight to the standing stage.

Between 10 and the 16 months

- *On average, a baby becomes a toddler, able to walk upright and unassisted.*

There is, however, considerable variation in timing from child to child.

Some are up and running by 12 months, while others take their time and do not learn to walk until up to two years after birth,

READING 2 (Cont.-7)

- *Whatever the case, in less than two years, the child has come a long way – from a helpless, almost immobile newborn baby to an active miniature adult.*

The attainment of the walking stage is matched by the development of the muscles and the nervous system that coordinates them.

A child can now explore under its own steam.

MILESTONES

- *The ages below are average times by which children usually achieve certain skills. Each child develops individually and may progress slightly earlier or later:*
- *6 weeks → smile*
- *10 weeks → roll over from a sideways position onto the back*
- *4-6 months → raised head and shoulders from a face down position*
→ sit up with some support
- *7 months → pass a toy from one hand to the other.*
- *8 months → try to feed him or herself with a spoon*

MILESTONES (Cont.-1)

- *9 months* → *rise to sitting position*
- *12 months* → *understand simple commands*
→ *stand unsupported for a second or two*
- *18 months* → *walk unaided*
- *2 years* → *achieve bowel control*
→ *stay dry during the day*
- *3 years* → *talk in simple sentences*
→ *stay dry during some nights*
- *4 years* → *get dressed and undressed (with a little help)*
- *5 years* → *draw a figure with separate body and limbs*

SOCIAL BEHAVIOUR

- ***Playing***

*Play is a child's work and toys are the tools of the trade. As much as they need food, warmth and love, children need **play to enlarge their understanding of the world around them.***

*Playthings are the stimuli that spur a child to intelligently explore his or her universe, and while it is important to provide your child with a variety of materials and activities, it is just as **essential to provide the space in which to enjoy them.***

PLAYING (Cont.-1)

- *Children **think and dream.***

A clamor of toys and games will rob them off the chance to focus all their attention on one thing at a time.

Give your children the space they need so that they can totally immerse themselves in the challenge of moulding clay castles or brushing Teddy's teeth.

*If playing with objects teaches children how the world works then **making friends and playing with other children helps them to know how they fit in.***

PLAYING (CONT.-2)

- *Awareness of another's existence is the first step toward learning to **consider other people's needs.***

*The child that misses out on this interaction may find it **difficult to relate to people later on** and may withdraw, becoming shy and solitary.*

*Some children are naturally reluctant to leave mommy to a new friend, and it is **important that they be gently encouraged and never pushed.***

SHARING

- *Toddlers under three years of age may not always play together in complete harmony but they will **begin to learn toleration and cooperation.***

*It is essential in life that we learn “**how to play the games**” and those seeds are sown in time when a child begins to share not only toys but him or herself with another child.*

Play then becomes a cooperative discipline with rules and terms of honour.

DISCIPLINE

- *Sensitive parents will use discipline in a positive sense to teach rather than to punish, this helping their child **balance his or her own needs in concert with other.** In this way, the parent uses discipline to define clearly what **is or is not acceptable behaviour** enabling the child to live happily and in fellowship with others.*

The socially healthy child learns to share joyfully and to cultivate the art of persuasion rather than force.

DISCIPLINE (Cont.)

- *The child who understands how to give the same attention to another child that he or she enjoys for him or herself **will never lack for friends.** Children whose parents inspire imaginative thought will be able to find their own way of expressing themselves, thus sharing not only their toys but their experiences of living.*
- *(Sumber: Canadian Medical Association: Complete Guide to Medical Symptoms)*

READING 3. THE GROWING CHILD

- ***From learning to walk to reaching adolescence, a child undergoes an amazing series of changes.***
- *By the end of the first dozen or so years, a child is on the verge of sexual maturity and is able to walk, speak, write and communicate socially with sophistication and fluency. Reaching this point from the helplessness of infancy seems to happen with little apparent effort. The child seem programmed to learn new things easily, and the early years are a time when fresh knowledge and skills are absorbed. Thereafter learning new skills becomes increasingly difficult.*

THE GROWING CHILD (Cont.-1)

- It is during the childhood years that human develop the skills for understanding and manipulating the world around them – a world of objects, activities, social interactions and the consequences of action.*

But in addition to the intellectual changes, there are physical changes.

The first 18 months or so of life see dramatic rates of growth; but these then slow down and body shapes alter more gradually.

THE GROWING CHILD (Cont.-2)

- *A stocky toddler becomes a taller five-year-old with longer limbs, making the child seem thinner.*

This thin look lasts until muscle growth catches up with the height gain.

In the middle phase of childhood, between five and seven years, the first set of teeth, or milk teeth, start to be replaced by the permanent adult set.

As the teenage years approach, there is another growth spurs. This mark the beginning of adolescence, the coming of sexual maturity.

READING 4: TIPS “PENGAMANAN”

A child’s newly developed mobility means that he or she is far more vulnerable to accidents than ever before.

His or her agility will most certainly exceed ability to sense danger and parents must take sensible precautions

(Pada saat mobilitasnya mulai berkembang, anak akan lebih mudah terkena cedera. Kecerdasan mentalnya akan melebihi perasaan terhadap bahaya, oleh karenanya para orang tua harus sangat berhati-hati menjaganya)

The Sitting Baby

Most babies from as early as 6 weeks like to be propped up so that they can look around.

*A baby seat is ideal, but the baby must be securely fastened and the chair placed on a **non-skid surface**.*

*If you prop your baby up in a regular chair or on the floor make sure he or she is surrounded by **plenty of pillows** so that he or she does not topple over.*

*Beware of **flimsy furniture** that the baby might pull over when trying to stand up.*

The Crawling Baby

A crawling baby must be carefully watched.

Make sure the area is clean smooth surfaced and free of loose wires, sharp-edged or rickety (goyang) furnishings and open fires.

Trailing tablecloths, low-hanging plants, breakables and the dog's water bowl should be removed.

Always have safety gates across stairways and don't let your child crawl anywhere near a door that may open.

Never have crawling baby unsupervised.

The Walking Baby

*Once your child begins walking **leave shoes and socks off as often as possible** as they can cause a baby to slip and fall.*

*Toddler who walk can reach and climb so **don't leave the windows wide open.***

*Banisters should be too narrow to squeeze through and **stairway fenced off.***

*Swinging or sliding doors should **be locked and locks must be out of reach.***

*Possible hiding places like cupboards, trunks and old kitchen appliances must be **securely fastened.***

Walking Baby (cont.)

*Turn all the **handles of pots and pans** away from the front of the stove or install a guard around it.*

*Never leave anything boiling or hot on a reachable surface, and make sure that **all sharp kitchen utensils** are stowed away.*

*Glass doors should have obvious markers and yard gates with road access must be **firmly shut**.*

*Lock all medicines well away. Never leave small **lying** around that can be shoved **into nose and ear** **never leave your child unsupervised.***

(Sumber: CMA: Complete Guide to Medical Symptoms)

READING 5. MILK TO FEED A BABY

- *Milk to feed a baby is made in gland cells in the breast under the influence of a hormone – **prolactin** – from the pituitary gland.*

*When a baby sucks on a nipple, this provokes the pituitary to release a surge of another hormone, **oxytocine**. This has the effect of making the smooth muscle in the breast contract, moving milk towards the nipple and releasing it as the baby needs it.*

- *The release of milk in response to the baby's sucking is the “**milk let-down**” reflex.*

Nourishment for the newborn

- *A new mother's breasts produced about **1 liter of milk a day**, but for up to three days after birth, the breasts give a **protein-rich liquid colostrum**.
This **contains antibodies**, to protect the baby from disease, and hormones that **stimulate the lining of its digestive tract** to digest milk.*
- *Milk's composition varies from mammal to mammal.*

READING 3. (Cont.-1)

- *Human milk has the same fat content but much less protein and more sugar (lactose) than cow's milk.*

Seal milk has more than 10 times the amount of fat and protein found in human milk.

This thick, rich milk enable a young seal to grow fast on its birth beach and lay down a thick layer of blubber before going to sea.

- (Sumber: Dr. Philip Whitfield: The Human Body Explained)

GANGGUAN PADA BAYI DI BAWAH 1 TAHUN

1. Penambahan berat badan lambat
2. Bergadang malam hari
3. Demam
4. Masalah kulit
5. Banyak menangis/cengeng
6. Masalah pemberian minum/makan
7. Muntah
8. Diare

GANGGUAN PADA MASA KANAK-KANAK

1. Rasa kurang sehat
2. Pertumbuhan lambat
3. Penambahan berat berat cepat
4. Masalah tidur
5. Mengantuk
6. Demam
7. Rasa gatal-gatal
8. Pingsan, pusing dan kejang
9. Sakit kepala
10. Lamban

GANGGUAN PADA MASA KANAK-KANAK (Lanjutan-1)

11. Bingung
12. Sulit berbicara
13. Masalah perilaku
14. Kesulitan di sekolah
15. Masalah rambut kepala dan kuku
16. Bentol-bentol dan ruam kulit
17. Ruam dengan demam
18. Masalah mata
19. Gangguan pengelihatatan (visus mata)
20. Sakit telinga

GANGGUAN PADA MASA KANAK-KANAK (Lanjutan-2)

21. Tuli
22. Rinorrhea dan nasal obstruction
23. Sakit tenggorokan
24. Batuk-batuk
25. Napas tersengal-sengal
26. Suara napas berisik
27. Sakit gigi
28. Muntah
29. Abdominal pain
30. Hilang nafsu makan

GANGGUAN PADA MASA KANAK-KANAK (Lanjutan-3)

31. konstipasi
32. Feces tidak normal
33. Masalah BAK
34. Masalah membiasakan menggunakan toilet
(ngompol)
35. Sakit lengan dan tungkai
36. Sakit persendian
37. Masalah kaki
38. Masalah genital wanita/laki-laki

(Sumber: CMA, Complete Guide To Medical Sysmtoms)

AUTISM

- Satu kondisi anak **gagal menjalin hubungan sosial dengan orang lain**. (Istilah autis dulu digunakan untuk sebutan bagi gangguan yang terjadi pada schizophrenia, saat ini sudah tidak digunakan lagi).

Insiden:

Autism termasuk yang jarang ditemukan, 2->4/10.000 anak. Laki 3x > banyak dari perempuan, > pada kelas sosial tinggi.

Umumnya terdiagnosis sejak 30 bulanan dan tanda-tanda jelas dalam tahun-tahun pertama perkembangan anak.

KAUSA

- Kausa persis belum dapat ditemukan.
- Bukti yang jelas berbasis fisik, $\frac{1}{4}$ autisme menunjukkan gangguan neurologis dan kejang epileptik timbul pada $\frac{1}{3}$ dewasa → memberi kesan adanya kerusakan otak.
- Teori kuno menyebut: akibat kurangnya kasih sayang dan kehangatan dalam keluarga → ini sekarang sudah dibantah.
- Umumnya, ortu bisa sampai stress menghadapi reaksi penolakan anak dengan gangguan terkait.

SIMTOMA DAN TANDA-TANDA

- Kadang dalam beberapa bulan-bula pertama normal kemudian tidak respons terhadap stimulus.
Tanda pertama bisa sebagai penolakkan bila ditimang, dan menangis keras bila dipeluk dan minta diturunkan dari gedongan, walau sakit atau lelah.
- Tidak menjalani hubungan mesra dengan anggota keluarga. Anak akan menghindari kontak mata, dan lebih senang main menyendiri, dan seringnya tidak hirau terhadap ortu atau perasaan orang lain atau norma sosial lain.

SIMTOMA DAN TANDA-TANDA (Lanjutan-1)

- Adanya resisten ekstrim untuk berubah adalah penampilan yang penting.
Anak akan menolak keras terhadap perubahan rutin atau gangguan terhadap aktivitasnya.
Bermain permainan ritual dan terkadang sangat terpukai terhadap obek yang tidak umum atau koleksi, atau terobsesi terhadap suatu topik atau ide tertentu.
Ketertarikan terhadap satu aktivitas yang sama sulit untuk anak autism dapat mengikuti petunjuk atau pelajaran yang lain

SIMTOMA DAN TANDA-TANDA (Lanjutan-2)

- Kemampuan berbicara lambat, kurang mampu mengerti atau meniru kata, atau isyarat, dan respons terhadap suara sangat lemah dan salah.
Suara bicara bisa seperti robot dan tidak mengucapkan suara kata yang diminta. Seringnya anak mengucapkan kata dan pantulan suara kata yang telah diucapkan.
- Berjalan dengan menjinjit-jinjit, flicking (kedap-kedip) and twiddling (memutar-mutarkan) jari-jari berjam-jam dan akhirnya bergoyong-goyang dan cedera, teriak keras mendadak, hiperaktivitas.
- Rasa takut yang luar biasa menyulitkan belajar aktivitas kerja manual

SIMTOMA DAN TANDA-TANDA (Lanjutan-3)

- Di samping kekurangan di atas, perkembangan fisik dan penampilan serta koordinasi otot normal.
Terkadang memiliki keterampilan aktifitas tersendiri → kemampuan di bidang penguasaan alat musik tertentu.
- Terapi:
Belum ditemukan yang efektif.
Sekolah luar biasa. Harus ada pendidikan bagi ortu.
Terapi prilaku untuk mengurangi kekerasan dan cedera diri.
Terapi obat hanya untuk gangguan tertentu:
epileptik atau hiperaktifitas.

OUTLOOK

- Bergantung pada IQ-nya, penguasaan berbahasa.
- Hanya 1/6 dapat mandiri
- Sisanya sangat ketergantungan
- Perlu institusi khusus untuk tinggal.

(Sumber AMA, Encyclopedia of Medicine)

Genetic Disorders

- Gangguan yang seluruh atau sebagian tubuh yang timbul akibat kesalahan yang ada di dalam material genetik sel yang diturunkan oleh orangtuanya, yakni: pada gen-gen yang terbentuk dari substansi DNA (deoxyribonucleic acid) yang membentuk kromosom di dalam sel sel seseorang.
- Banyak penyakit timbul sebagian atau seluruhnya akibat penyebab genetik.

(Sumber AMA, Encyclopedia of Medicine)

Genetic Disorders (Lanjutan)

- Banyak gangguan genetik adalah kongenital. Namun demikian, genetik bukan sinonim dari kongenital.

Banyak defek genetik belum nampak saat dilahirkan dan baru nampak setelah beberapa tahun kemudian, banyak gangguan abnormalitas kongenital bukan genetik.

Gangguan genetik seringnya familial.

Adakalanya seorang anak lahir tidak terduga menderita gangguan genetik, padahal di dalam keluarga tidak ada riwayat gangguan genetik.

Beberapa Mekanisme terjadinya Gangguan Genetik

- Di dalam sel gen berfungsi mengontrol manufaktur enzyme dan protein lain-lian, yang berperan penting dalam sel dan tubuh secara keseluruhan.

Apabila material genetiknya defektif, maka bisa terproduksi protein-protein yang abnormal yang akan mengubah susunan kimia tubuh sehingga tubuh menjadi sakit.

Beberapa Mekanisme ... (Lanjutan)

- Gangguan genetik bisa muncul apabila material genetik yang abnormal terkiat ada di dalam sel individu yang bersangkutan. Ini hanya mungkin apabila di dalam sel telur atau sperm memang ada material genetik yang tidak normal.

Ada 2 (dua) jalan yang bisa terjadi:

1. satu atau kedua orang tuanya mengandung material genetik yang defektif;
2. ada tetjadi mutasi (perubahan ,material genetik) saat formasi sel telur atau sperma.

MUTASI

- Adalah satu di antara mekanisme yang mengakibatkan anak dari keluarga yang tidak memiliki riwayat gangguan genetik terlahir terkena gangguan genetik.

Contoh:

Pada gangguan genetik hemophilia, 1/3 kasus hemophilia terjadi akibat mutasi.

Hemophilia adalah penyakit gangguan perdarahan yang diturunkan, akibat kekurangan protein darah tertentu (biasanya pasien adalah laki-laki).

Hemophilia dengan HIV antibodi (+) tidak boleh mempunyai anak, hemophilia dengan HIV antibodi (-) harus counseling.

MUTASI (Lanjutan)

- Pada Mutasi terjadi suatu perubahan pada material genetik (DNA, yang merupakan bagian dari kromosom sel yang mengkode instruksi aktivitas sel)
- Banyak mutasi adalah netral atau tidak berbahaya bagi pasiennya; sebagian mutasi sangat jahat, bisa menimbulkan:
 - kanker,
 - cacat lahir, dan
 - penyakit hereditas.

Sangat jarang bahwa mutasi menguntungkan.

3 (three) Broad Classes of Genetic disorders

(1) Chromosome abnormalities

A child is born with an abnormal number of whole chromosomes, or extra or missing bits of chromosomes, in the cells, since chromosomes contain many genes, this can lead to multiple disturbances and disorders.

1/200 lahir hidup memiliki abnormalitas kromosom.

1/3 Abortus spontan memiliki abnormalitas kromosom

Ini memberi gambaran bahwa adanya abnormalitas kromosom mengakibatkan fetus tidak kompatibel untuk bisa hidup di luar

3 (three) Broad Classes of ...(Cont.-1)

(2) Unifactoral defects

Unifactoral disorders are caused by single defective gene or pair of genes; these disorders are distributed among the members of an affected family according to relatively simple laws of inheritance.

Unifactoral disorders bisa:

- Autosomal dominant*
- Autosomal recessive*
- X-linked recessive*

3 (three) Broad Classes of ... (Cont.-2)

(3) Multifactorial defects

Multifactorial disorders are caused by additive effects of several genes, along with environmental factors; the pattern of inheritance is less straightforward.

Disorders fall into this category:

- asthma,*
- IDDM,*
- schizophrenia,*
- clubfoot*
- cleft lip or palate.*

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CHROMOSOMAL ABNORMALITIES

Variations from normal in the number or structure of chromosomes contained within a person's cells.

In most cases, the chromosomal abnormality is present in all the cells, it may have anything from a lethal to virtually no effect, depending on the particular type of abnormality.

INCIDENCE & CAUSES

About 1 in every 200 babies born alive has a chromosomal abnormality.

Among spontaneously aborted fetuses, 1 in 3 has such an abnormality, this suggests that most chromosome abnormalities are incompatible with life and that those seen in babies born alive are actually the less serious ones.

INCIDENCE & CAUSES (Cont.)

The cause in most cases is some fault in the process of chromosomal division, either during the formation of the egg or sperm from which a person is derived, or during the first few divisions of the fertilized egg.

Occasionally, one of the parents has an abnormal arrangement of his or her chromosomes.

TYPES

A complete extra set of chromosomes per cell is called polyploidy and is lethal.

Other abnormalities can be classified according to whether they involve the 44 autosomes or the two sex chromosomes .

Those affecting the autosomes are slightly less common than sex chromosome abnormalities, but tend to produce more serious and widespread effects.

AUTOSOMAL ABNORMALITIES

Extra autosome means that one of the 22 pairs of autosomes occurs in triplicate instead of as a pair – a phenomenon called trisomy (Down's Syndrome).

Sometimes, a part of a chromosome is missing (cru du chat syndrome) or an extra bit is present and joined to another chromosome.

All these autosomal abnormalities as well as others causing physical defects of varying severity, tend to cause mental retardation as well.

AUTOSOMAL ABNORMALITIES (Cont.-)

*Occasionally, a person has a normal chromosomal complement, but part of one autosome is not in its proper position: rather, it is joined to another chromosome – a phenomenon called **translocation**.*

The person is normal, but some of his or her children may suffer from an abnormality.

SEX CHROMOSOME ABNORMALITIES

*About one girl in 2.500 is born with only one X chromosome in her cells instead of 2 – a condition known as **Turner's syndrome**.*

The annotation for this is 45 XO, meaning 45 instead of 46 chromosomes, with just a single X chromosome.

The condition causes characteristic physical abnormalities.

Defective female sexual development, and infertility.

SEX CHROMOSOME ABNORMALITIES (Cont.-1)

All other sex chromosome abnormalities involve extra chromosomes.

A boy born with one or more extra X chromosome has Klinefelter's syndrome. The annotation for this is usually 47 XXY OR 48XXX (1/500 male births), although it is often not diagnosed until puberty. The condition causes defective male sexual development infertility, and, in some cases, mental retardation.

SEX CHROMOSOME ABNORMALITIES (Cont.-2)

Some women are born with extra X chromosome (47XXX) and men with an extra Y (47XYY).

These people are usually normal physically, but may have an increased risk of mental retardation and perhaps psychological problems.

The presence of the extra chromosome is recognized only if a special attempt is made to discover it.

DIAGNOSIS & TREATMENT

- *Abnormalities are diagnosed by a chromosome analysis, which is now possible early in pregnancy using chorionic villi sampling.*

Because of the fundamental nature of these defects – affecting every one of a person’s cells – no “cure” is possible.

Most babies with autosomal chromosomal defects, apart from those with Down’s syndrome, do not survive long. Hormonal and surgical treatment can help correct some of the development defects characteristic of Klinefelter’s and Turner’s syndromes.

DIAGNOSIS & TREATMENT (Cont.)

Anyone with a child or other member of the family with chromosomal abnormality should obtain genetic counselling to establish the risk of his or her prospective children being affected and also to discuss other consideration of family planning.

DOWN SYNDROME

- Suatu kondisi abnormalitas kromosom yang menghasilkan anak handicap (rintangan) mental disertai penampilan wajah khas (dahulu dikenal dengan istilah mongoloid).
- **Kausa:**
Mysterious sampai tahun 1959. Sekarang diketahui bahwa setiap sel tubuh Down syndrome memiliki kromosom 47, > satu pada kromosom 21, akibat pada stadium pertama formasi sel sperma atau sel telur, gagalnya kedua kromosom 21 dari orang tuanya menjadi dua daughter (anak) cells.
→ ada telur dan sperma yang mengandung jumlah ekstra kromosom 21.

KAUSA (Lanjutan)

- Kejadian tersebut lebih banyak pada bumil usia lebih 35 tahun, sehingga disimpulkan umumnya defektif ada pada sel telur ketimbang pada sel sperma.
- Kausa lain yang lebih kecil adalah adanya defek pada kedua sel ortu → ***translocation***, sebagian kromosom 21 dari ortu bersatu dengan kromosom lain. Ortu tidak terkena gangguan namun berisiko menghasilkan keturunan Down's syndrom

INSIDEN & SIMTOMA

- **Insiden:** 1/650 bayi lahir dengan Down Syndrome. Insiden terpengaruhnya janin meningkat tajam s/d $\frac{1}{4}$ pada bumil di atas 40 th. Risiko juga bisa terjadi pada bumil diatas 35th dengan riwayat terkait di keluarga → dilakukan analisis kromosom melalui amniocentesis atau chorionic villus sampling. → di USA boleh dilakukan terminasi kehamilannya.

Simtoma: Mata menurun di luar lipatan kulit hidung, tulang hidung tidak menonjol, ukuran mukanya kecil, lidah besar menjurus ke luar , kepala belakang gepeng, tangan pendek namun lebar, garis garis telapak tangan lurus sejajar (tidak mirip huruf M).

SIMTOMA (Lanjutan)

- IQ antara 30 – 80, ada yang sampai bisa baca dan belajar. Sekolah di sekolah khusus.
Umumnya mereka sangat ramah dan penyayang, dan berperangai gembira, sangat mudah bergaul dengan anggota keluarga lainnya.
- $\frac{1}{4}$ terkena defek jantung kongenital, atresia intestinum, defek pendengaran, dan leukaemia akut, mudah terkena infeksi.
Pada Down Syndrome dewasa mudah terserang arteriosclerosis.

DIAGNOSIS & TERAPI

- Karena penampilan yang khas maka Down Syndrome mudah terdeteksi sejak lahir. Diagnosis atas dasar ditemukannya jumlah kromosom di sel leukosit.

Terapi:

- Tidak ada terapi khusus
- Pendidikan harus di sekolah khusus
- Anak lebih senang tinggal pada keluarga
- Tampilan wajah bisa dikoreksi dengan operasi plastik

Outlook:

Tidak banyak yang mencapai usia di atas 10 th. (akibat defek kongenital)

Kemajuan teknologi medis → sampai usia pertengahan.