20 Genetika Klinis

Waktu

Pencapaian kompetensi

Sesi di dalam kelas : 4 x 50 menit (*classroom session*)
Sesi dengan fasilitasi pembimbing : 4 x 100 menit (*coaching session*)
Sesi praktik dan pencapaian kompetensi : 4 minggu (*facilitation and assessment*)

Tujuan pembelajaran umum

Setelah mengikuti sesi ini, peserta didik mampu untuk:

1. mengidentifikasi pasien atau keluarga yang memerlukan pelayanan genetik,
2. membantu mereka mencapai fasilitas genetik
3. menjawab pertanyaan selama proses konsultasi genetik

Tujuan pembelajaran khusus

Setelah mengikuti sesi ini peserta didik akan:

1. Differentiate between chromosomal, single gene, and multifactorial disorders.
2. Describe what might cause nontraditional patterns of inheritance.
3. Identify the influence that new mutations and susceptibility genes have on general health and well being.
4. Describe the basis for, and significance of, genetic tests. Obtain and analyze a family history to determine if the family should be referred for genetic services.
5. Define the principles of teratology.
6. Identify women at increased risk of having children with birth defects or genetic disorders.
7. Recognize unusual characteristics that suggest a genetic abnormality
8. Define what observations you might make during the course of a person's life that would lead you to conclude that a genetic consultation is appropriate. (Genetics and life cycle)
9. Provide families with specific information about what to expect when they are referred for routine prenatal diagnostic procedures or clinical genetic services
10. Describe some of the unique ethical, legal and social issues that may arise when providing genetic services
11. Principles of pre- and postnatal genetic screening
12. Recognize the interrelationship between genetic disorders, family values, culture and family dynamics.
13. Define the grief process and how this might affect the referral process
    a. Difficulties in coping
    b. Difficulties in making choice
14. Identify your own personal values and the influence they have on the patient-provider interaction.

1. Therapeutic abortion
2. Euthanasia
3. How to be non-directive as a counselor

Strategi pembelajaran

Tujuan 1.

1. Mampu membedakan kelainan kromosomal, *single gene*, atau *multifactorial disorders*.
2. Mampu menjelaskan cara penurunan non tradisional.
3. Mengidentifikasi pengaruh mutasi genetik pada terhadap kesehatan manusia

Untuk mencapai tujuan ini maka dipilih metode pembelajaran sbb:

- *Interactive lecture*
- *Small group discussion (journal reading,Case study, Problem based learning,etc)*
- *Peer assisted learning (PAL)*
- *Bedside teaching*
- Praktek mandiri dengan pasien

Must to know:

- *Chromosomes*
- *Chromosomal Patterns of Inheritance*
- *Abnormalities in Chromosome Structure*
- *Mendelian Patterns of Inheritance*
- *Multifactorial Inheritance*
- *Nontraditional Patterns of Inheritance*
- *Testing for Genetic Disorders*
- *DNA Molecular Diagnosis*

Tujuan 2.

1. *Obtain and analyze a family history to determine if the family should be referred for genetic services.*
2. *Define the principles of teratology.*
3. *Identify women at increased risk of having children with birth defects or genetic disorders.*
4. *Recognize unusual characteristics that suggest a genetic abnormality.*
5. *Define what observations you might make during the course of a person's life that would lead you to conclude that a genetic consultation is appropriate. (Genetics and life cycle)*

Untuk mencapai tujuan ini maka dipilih metode pembelajaran sbb:

- *Interactive lecture*
- *Small group discussion (journal reading,Case study, Problem based learning,etc)*
- *Peer assisted learning (PAL)*
- *Bedside teaching*
- Praktek mandiri dengan pasien

Must to know:

- *Taking a Family History*
- *Patterns of Inheritance: Analyzing a Family History*
- *Teratogens*
- *Preconception and Prenatal Risk Assessment Tools*
- *Basic Dysmorphology*
- *Age dependency, epigenetics*

Tujuan 3.

1. *Provide families with specific information about what to expect when they are referred for routine prenatal diagnostic procedures or clinical genetic services.*
2. *Describe some of the unique ethical, legal and social issues that may arise when providing genetic services.*
3. *Principles of pre- and postnatal genetic screening*

Untuk mencapai tujuan ini maka dipilih metode pembelajaran sbb:

- *Interactive lecture*
- *Small group discussion (journal reading,Case study, Problem based learning,etc)*
- *Peer assisted learning (PAL)*
- *Bedside teaching*
- Praktek mandiri dengan pasien

Must to know:

1. Prenatal genetic services:
   a. Prenatal diagnosis
   b. Prenatal screening
   c. Neonatal screening
2. General genetic sevices:
   a. Predictive genetic testing
3. Ethical and social implications of genetic disorders
   a. How to involve family members
   b. Insurance issues
   c. Employment issues

Tujuan 4.

1. *Recognize the interrelationship between genetic disorders, family values, culture and family dynamics.*
2. *Define the grief process and how this might affect the referral process*
   a. *Difficulties in coping*
   b. *Difficulties in making choice*
3. *Identify your own personal values and the influence they have on the patient-provider interaction.*
   a. *Therapeutic abortion*
   b. *Euthanasia*
   c. *How to be non-directive as a counselor*

Untuk mencapai tujuan ini maka dipilih metode pembelajaran sbb:

- *Interactive lecture*
- *Small group discussion (journal reading,Case study, Problem based learning,etc)*
- *Peer assisted learning (PAL)*
- *Bedside teaching*
- Praktek mandiri dengan pasien

Must to know:

1. *The Referral Process*
2. *Family Dynamics*
3. *The Grief Process*
    - *The four dimensions of the mourning process are (1) shock and numbness, (2) yearning and searching, (3) disorientation and disorganization, and (4) resolution and reorganization. The four dimensions do not follow a set order and a person may experience feelings from several stages at one time.*
    - *Implications and Interventions for Professionals recommendations offer when working with the parent of a child with a congenital defect or a person with a newly diagnosed genetic condition*
4. *Organizational Barriers*
5. *The Interview*
6. *Values Clarification*

Persiapan sesi :

- Materi sesi dalam program *power point:*

  Genetika Klinis
  Slide
  1. Chromosomes
  2. Chromosomal Patterns of Inheritance
  3. Abnormalities in Chromosome Structure
  4. Mendelian Patterns of Inheritance
  5. Multifactorial Inheritance
  6. Nontraditional Patterns of Inheritance
  7. Testing for Genetic Disorders
  8. DNA Molecular Diagnosis Taking a Family History
  9. Patterns of Inheritance: Analyzing a Family History
  10. Teratogens
  11. Preconception and Prenatal Risk Assessment Tools
  12. Basic Dysmorphology
  13. Age dependency, epigenetics
  14. Prenatal Genetic Services :
  15. General Genetic Services
  16. Ethical and Social Implications of Genetic Disorders
  17. The Referral Process
  18. Family Dynamics

19. The Grief Process
20. Organizational Barriers
21. The Interview
22. Values Clarification

- Sarana dan alat Bantu :
  - Penuntun belajar (*learning guide*)
  - Tempat belajar (*training setting*): Rawat jalan dan rawat inap
  - Audiovisual

Kepustakaan

1. The Metabolic and Molecular Bases of Inherited Disease, 4 volume set by Charles R. Scriver (Editor), William S. Sly (Editor), Barton Childs, Arthur L. Beaudet, David Valle, Kenneth W. Kinzler, Bert Vogelstein
2. Smith's Recognizable Patterns Of Human Malformation Sixth Edition (Smith's Recognizable Patterns of Human Malformation) by Kenneth Jones
3. Practical Genetic Counselling (A Hodder Arnold Publication) by Peter S. Harper

Kompetensi

……

Gambaran Umum

GENETIKA KLINIS

……

Contoh Kasus

STUDI KASUS
GENETIKA KLINIS

Arahan

Baca dan lakukan analisis terhadap studi kasus secara perorangan. Apabila peserta lain dalam kelompok sudah selesai membaca contoh kasus, jawab pertanyaan yang diberikan. Gunakan langkah dalam pengambilan keputusan klinik pada saat memberikan jawaban. Kelompok yang lain dalam ruangan bekerja dengan kasus yang sama atau serupa. Setelah semua kelompok selesai, dilakukan diskusi studi kasus dan jawaban yang dikerjakan oleh masing-masing kelompok.

Studi Kasus 1

Seorang bayi perempuan usia 8 bulan dibawa oleh orangtuanya ke RS karena sampai saat ini belum dapat duduk, selain itu juga sejak lahir jika menyusu hanya sebentar-sebentar. Pada pemeriksaan fisis tidak didapatkan *facial dysmorphism* tetapi kepala masih terkulai kesannya aksial hipotonia. Pada pemeriksaan jantung didapatkan kardiomiopati. Kedua orang tua tidak ada pertalian darah, kedua kakak lelaki pasien berusia 5 dan 3 tahun sejauh ini tampak normal. Pemeriksaan laboratorium menunjukkan adanya defisiensi enzim *acid maltase*.

Apa penyakit anak ini ?

Apa dasar diagnosisnya secara klinis dan laboratoris ?

Termasuk kelompok penyakit apa ?
Bagaimana patogenesisnya ?
Apakah ada pengobatannya ?
Perlukan *genetic counselling* pada pasien ini, bagaimana ?

Jawaban:

Studi Kasus 2

Seorang anak lelaki berusia 9 tahun dibawa berobat oleh orang tuanya karena terlalu gemuk. BB saat ini adalah 100 kg dengan TB 128 cm. Anak ini dilahirkan prematur 36 minggu dengan BL 2,4 kg, sampai usia 1 tahun BB-nya sulit naik tetapi setelah itu nafsu makannya mendadak berlebihan sehingga pada usia 3 tahun beratnya 30 kg. Pasien juga mengalami keterlambatan perkembangan karena baru dapat duduk sendiri pada usia 1,5 tahun dan berjalan sendiri usia 2,5 tahun. Saat ini dia duduk di kelas 1 SD tetapi sulit mengikuti pelajaran sehungga 2 kali tidak naik kelas dan pada pemeiksaan psikologi didapatkan adanya IQ 60. Anak ini merupakan anak pertama dari pasangan orang tua yang *non consanguinitas*. Pada pemeriksaan fisik didapatkan adanya *undescensus testiculorum* bilateral, tangan dan kaki kecil, mukanya khas suatu sindrom. Pemeriksaan analisis kromosom menunjukkan delesi kromosom 15.

Apakah penyakit yang diderita anak ini?
Patogenesis sindrom tersebut ?
Bagaimana tatalaksana sindrom ini ?
Genetic couselling untuk pasien ini ?

Jawaban:

Tujuan pembelajaran

Proses, materi dan metoda pembelajaran yang telah disiapkan bertujuan untuk meningkatkan pengetahuan, keterampilan, dan perilaku yang terkait dengan pencapaian kompetensi dan keterampilan yang diperlukan dalam mengenali dan menatalaksana genetika klinis yaitu :

1. Differentiate between chromosomal, single gene, and multifactorial disorders.
2. Describe what might cause nontraditional patterns of inheritance.
3. Identify the influence that new mutations and susceptibility genes have on general health and well being.
4. Describe the basis for, and significance of, genetic tests. Obtain and analyze a family history to determine if the family should be referred for genetic services.
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    1. Therapeutic abortion
    2. Euthanasia
    3. How to be non-directive as a counselor

Evaluasi

- Pada awal pertemuan dilaksanakan pre-test yang bertujuan untuk menilai kinerja awal yang dimiliki peserta didik dan untuk mengidentifikasi kekurangan yang ada.
- Selanjutnya dilakukan "small group discussion" bersama dengan fasilitator untuk membahas kekurangan yang teridentifikasi, membahas isi dan hal-hal yang berkenaan dengan penuntun belajar, kesempatan yang akan diperoleh pada saat bedside teaching dan proses penilaian.
- Setelah mempelajari penuntun belajar ini, mahasiswa diwajibkan untuk mengaplikasikan langkah-langkah yang tertera dalam penuntun belajar dalam bentuk *role-play* dengan teman-temannya (*peer assisted learning*) atau kepada SP (*standardized patient*). Pada saat tersebut, yang bersangkutan tidak diperkenankan membawa tuntunan belajar, tuntunan belajar dipegang oleh teman-temannya untuk melakukan evaluasi (*peer assisted evaluation*). Setelah dianggap memadai, melalui metoda *bedside teaching* di bawah pengawasan fasilitator, peserta didik mengaplikasikan penuntun belajar kepada pasien sesungguhnya. Pada saat pelaksa-naan, evaluator melakukan pengawasan langsung (*direct observation*), dan mengisi formulir penilaian sebagai berikut:
  - **Perlu perbaikan:** pelaksanaan belum benar atau sebagian langkah tidak dilaksanakan
  - **Cukup:** pelaksanaan sudah benar tetapi tidak efisien, misal pemeriksaan terlalu lama atau kurang memberi kenyamanan kepada pasien
  - **Baik:** pelaksanaan benar dan baik (efisien)
- Setelah selesai *bedside teaching*, dilakukan kembali diskusi untuk mendapatkan penjelasan dari berbagai hal yang tidak memungkinkan dibicarakan di depan pasien, dan memberi masukan untuk memperbaiki kekurangan yang ditemukan.
- Self assessment dan *Peer Assisted Evaluation* dengan mempergunakan penuntun belajar
- Pendidik/fasilitas:
  - Pengamatan langsung dengan memakai *evaluation checklist form* (terlampir)
  - Penjelasan lisan dari peserta didik/ diskusi
  - Kriteria penilaian keseluruhan: cakap/ tidak cakap/ lalai.
  - Di akhir penilaian peserta didik diberi masukan dan bila diperlukan diberi tugas yang dapat memperbaiki kinerja (*task-based medical education*)
- Pencapaian pembelajaran: pencapaian tingkat kompetensi A2-B2-C2

Instrumen penilaian

- **Kuesioner awal (MCQ / esei):**

PRACTICE ACTIVITY 1

1. What is the difference between a chromosome abnormality and a genetic disorder?
2. Chromosome abnormalities account for what percent of miscarriages?
3. Describe meiosis.
4. Describe nondisjunction.

PRACTICE ACTIVITY 2

Use a **T** or **F** to show whether each statement is **true** or **false**.

1. All parents of children with chromosome abnormalities should have a chromosome study.
2. Individuals who carry a balanced chromosome rearrangement cannot have normal children.

PRACTICE ACTIVITY 3

Define the following terms:

1. gene
2. homozygote
3. heterozygote
4. genotype
5. phenotype
6. nonpenetrance
7. expressivity

Use a **T** or **F** to show whether each statement is **true** or **false**.

8. It is possible to diagnoses PKU by doing a routine chromosome study.
9. The parents of a child with PKU will be physically and intellectually normal.
10. A child with an autosomal dominant single gene disorder will always have at least one affected parent.
11. If the first child born to a person with Marfan syndrome is affected, his next child will be unaffected.
12. Males with hemophilia have a 50% chance of having a son with hemophilia.
13. Women will never develop X-linked recessive single gene disorders.

PRACTICE ACTIVITY 4

Define the following terms:

1. imprinting
2. contiguous gene syndromes
3. triplet repeats

PRACTICE ACTIVITY 5

1. List 3 reproductive choices available to couples who carry genes coding for Tay-Sachs disease.
2. List 2 possible drawbacks to presymptomatic testing for Huntington disease.

PRACTICE ACTIVITY 6

Use a **T** or **F** to show whether each statement is **true** or **false**.

1. Every Caucasian person should be offered carrier testing for the CF gene.
2. If the exact gene mutation is not known, presymptomatic testing is not possible.

ANSWERS:

PRACTICE ACTIVITY 1: ANSWERS

1. A chromosome abnormality is caused by the presence of extra or missing chromosome material. The genes in a person with a chromosome abnormality are normal. It is the number of genes (increased or decreased) that is abnormal. A genetic disorder is caused by a change in a single gene, or genetic message, coding for a particular trait.
2. 50-60%
3. Meiosis is a special process of cell division which results in the formation of eggs and sperm. During meiosis, the pairs of chromosomes are replicated and separated, and the resulting gametes contain a set of 23 chromosomes.
4. Nondisjunction occurs when the normal separation of the chromosome pairs during meiosis is disrupted. If nondisjunction occurs, gametes are formed that contain too many or too few chromosomes.

PRACTICE ACTIVITY 2: ANSWERS

1. **False** Chromosome studies on parents should be ordered if a child is found to have a structural chromosome abnormality (e.g., translocation, deletion, inversion, etc.) to rule out carrier status. However, aneuploidy such as trisomy 21 and monosomy X (Turner syndrome), is caused by nondisjunction. As nondisjunction occurs sporadically at the time the egg or sperm is formed, it is assumed that the parents of these children have a normal chromosome complement.
2. **False** While it is true that individuals who carry a balanced chromosome rearrangement are more likely to have miscarriages and children with chromosome abnormalities, the vast majority can have chromosomally normal children. One exception to this rule is a person who carries a 21;21 translocation. He/she will either pass on the 21;21 translocation chromosome and have a child with Down syndrome, or he/she will fail to pass on the 21;21 translocation chromosome and the fetus, with monosomy 21, will be miscarried. Thus, a 21;21 translocation carrier has a 100% chance of having a child with Down syndrome.

PRACTICE ACTIVITY 3: ANSWERS

1. A **gene** is a submicroscopic segment of DNA that codes for the synthesis of a protein.
2. A **homozygote** is a person who has a pair of similar genes (or alleles) at a particular locus or site.
3. A **heterozygote** is a person who has two different genes (or alleles) at a particular location on a chromosome.
4. **Genotype** refers to the pair of genes (or alleles) a person inherits that code for a particular trait.
5. **Phenotype** refers to a person's observable physical, biochemical or physiological characteristics. A person's phenotype is determined by the interaction between genes and the environment. One phenotype may be due to different genotypes, as was described in the PKU example.

6. When an individual who inherits a gene coding for an abnormal dominant trait is phenotypically normal, the trait is said to be **nonpenetrant**.
7. **Expressivity** refers to the severity of a particular genetic disorder. In the case of some autosomal dominant single gene disorders, a parent may have only a few subtle characteristics that are suggestive of the disorder, whereas the child may have more obvious physical features. This would be an example of variable expressivity.
8. False PKU is caused by a change in a single gene. As each chromosome is made up of thousands of genes, it is impossible to distinguish one gene from another on chromosome study. A child with PKU would have a normal karyotype.
9. True While it is true that the parents of a child with PKU carry the abnormal gene coding for this condition, in the majority of cases, they will also carry the normal gene coding for PAH production. Therefore, they are able to break down phenylalanine and will have no obvious signs of phenylketonuria.
10. False A dominant disorder may be caused by a new mutation, or gonadal mosaicism. It is also possible that one of the parents is a nonpenetrant carrier, or has subtle signs of the disorder and is considered unaffected.
11. False Each pregnancy is an independent event. Given that each gene pair separates in meiosis, we know that 50% of his sperm contain the gene coding for Marfan syndrome and 50% of his sperm contain the gene coding for normal development. Whether his next child is affected or not depends on which gene is present in the sperm that fertilizes the egg.
12. False To have a son, a man must pass on his Y chromosome. Therefore, male children born to men with hemophilia will not inherit the X-linked gene coding for this condition.
13. False While unlikely, some women who carry a gene coding for an X-linked recessive genetic condition may develop signs of this condition, due to unequal X inactivation of the normal X chromosome. Women who inherit only one X chromosome may also have an X-linked disorder, such as Duchenne muscular dystrophy, in addition to Turner syndrome.

SUMMARY

Key points to remember about multifactorial inheritance:

- *There is a similar risk for first degree relatives (offspring, sibs or parents).*
- *Identical twins are not 100% concordant, indicating that there are nongenetic factors involved.*
- *The greater the number of affected relatives, the higher the recurrence risk. Empiric risk figures are used.*
- *The severity of the disorder and occasionally the sex of the affected individual may modify the risk.*

Some of the most common chronic diseases are multifactorial genetic disorders. Conditions with multifactorial inheritance include many birth defects, cancers, coronary artery disease, diabetes, hypertension, and mental disorders. It explains the familial distribution of many disorders. In general, the recurrence risk is based on experience and on observation of these disorders in the general population.

PRACTICE ACTIVITY 4: ANSWERS

1. **Imprinting** is a phenomenon whereby genes or chromosome segments are modified during meiosis. The imprinting process differs in males and females. Both maternal and paternal genes are necessary for normal development.
2. **Contiguous gene syndromes** refer to concurrent syndromes that occur due to duplication

or deletion of a series of genes that lie next to one another on a chromosome.

3. **Triplet repeats** are a sequence of three base pairs that occur in varying numbers in front of, within or just after a gene. The greater the number of repeated sequences, the more unstable the chromosome segment becomes during meiosis. If the number of repeats increases significantly, it may interfere with the normal gene function.

PRACTICE ACTIVITY 5: ANSWERS

1. The list may include any of the following: not having children, adoption, choosing to have children and taking the chance they will be affected, opting for prenatal diagnosis and terminating a pregnancy with an affected fetus, opting for prenatal diagnosis and preparing to deliver at a hospital where immediate treatment is available, or artificial insemination with a normal donor egg or sperm.
2. Possible drawbacks to presymptomatic testing for HD include depression because there are no known intervention strategies or cures; possible loss of employment or insurance; loss of hope for a family if the test is positive and the person chooses not to risk passing on the gene; survivor guilt, experienced by the sibs whose test turned out negative; etc.

PRACTICE ACTIVITY 6: ANSWERS

1. **False** Within the CF gene, there are hundreds of mutations that can result in the production of a nonfunctional protein. It is impractical to run tests looking for all of these mutations; therefore, CF carrier screening is usually reserved for those persons who have a family history of CF and their partners.
2. **False** If there are markers close to or within the gene in question, it may be possible to test affected and unaffected family members and determine which markers are traveling with or linked to the disease gene in a particular family. Once this is known, presymptomatic testing can be offered to at-risk family members.

- **Kuesioner tengah (MCQ / esei) :**

LESSON 1 POSTTEST

Write the karyotype for the following situations:

1. Normal female with a balanced Robertsonian translocation between chromosomes 13 and 21
2. Normal male with a balanced translocation between chromosome 10 and chromosome 7 with breakpoints on the long arm of 10 at band 22 and on the short arm of 7 at band 12
3. Down syndrome boy
4. Turner syndrome girl
5. Normal female with an inverted segment of chromosome 10 with breakpoints on the short arm at band 12 and on the long arm at band 22

Use a **T** or **F** to indicate whether each statement is **true** or **false**.

6. The incidence of Down syndrome rises with increasing maternal age.
7. Blood should be received in the genetics laboratory within 24 to 48 hours of sampling.

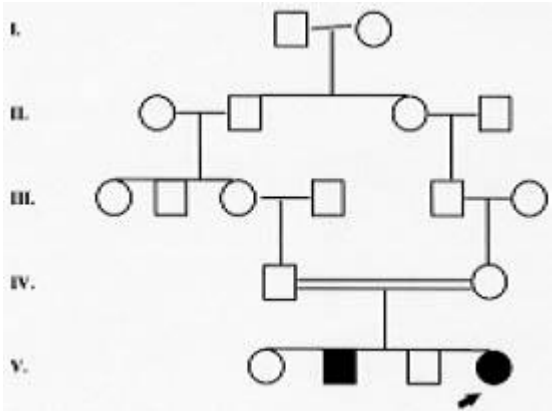
Indicate what type of sample should be sent for a chromosome study in the following situations:

8. To diagnose chromosome abnormality in the patient
9. To rule out mosaicism
10. To rule out a chromosome abnormality as the cause of a miscarriage
11. To rule out a fetal chromosome abnormality in pregnant older mothers

12. To diagnose leukemia/cancer

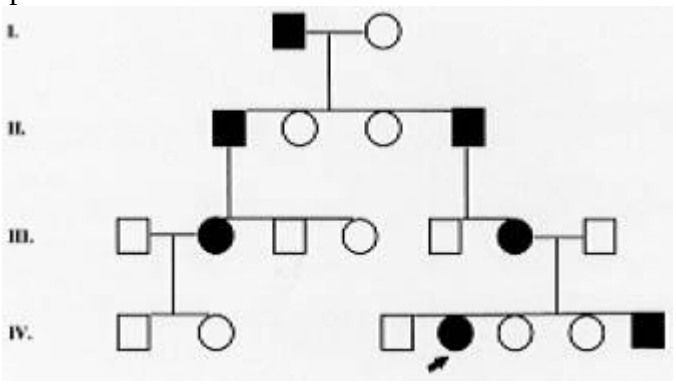
LESSON 2 POSTTEST

1. What is the first thing you do when collecting family history information?
2. List at least three things you should know about each member of your patient's family to include on a pedigree.
3. Why is it important to know your patient's ethnic background?
4. Why should you ask about consanguinity?
5. Analyze the following pedigree and state the most likely inheritance pattern, your reason for the choice and an example of the disease/condition.



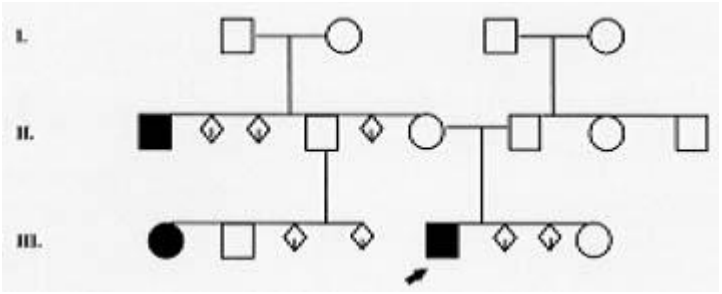
Pattern of Inheritance
Rationale
Example

6. Analyze the following pedigree and state the most likely inheritance pattern, your reason for the choice and an example of the disease/condition.



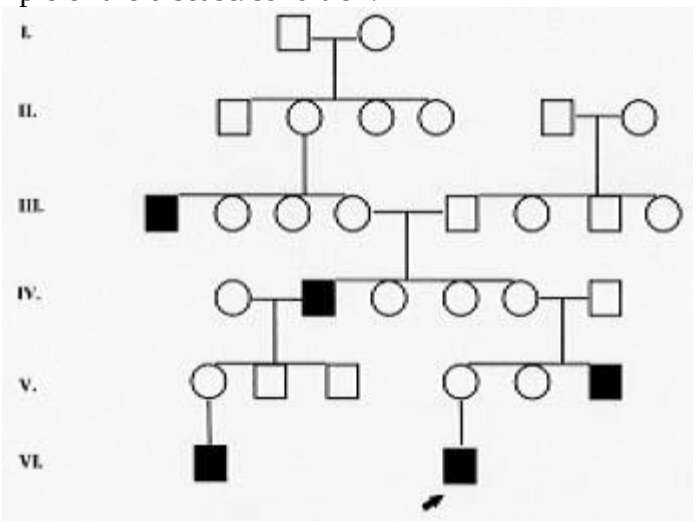
Pattern of Inheritance
Rationale
Example

7. Analyze the following pedigree and state the most likely inheritance pattern, your reason for the choice and an example of the disease/condition.



Pattern of Inheritance
Rationale
Example

8. Analyze the following pedigree and state the most likely inheritance pattern, your reason for the choice and an example of the disease/condition.



Pattern of Inheritance
Rationale
Example

9. List three factors that determine what effect prenatal exposure to a teratogenic agent will have on fetal development.

10. List two questions you can add to your existing preconception/prenatal tools that will assist you in the identification of couples who might benefit from a genetic evaluation or counseling.

Use a **T** or **F** to show whether each statement is **true** or **false**.

11. A dominant trait has a 25% risk of recurrence.
12. A recessive trait has a 50% risk of recurrence.
13. A child with short stature, mental retardation and dysmorphic features should be referred for genetic counseling.
14. Couples who have had three or more pregnancy losses should be referred for genetic counseling.

LESSON 3 POSTTEST

Use a **T** or **F** to show whether each statement is **true** or **false**.

1. Genetic counselors inform their patients of the appropriate course of action or reproductive choices they should make.

2. Every pregnant woman should have MSAFP or triple screening.
3. Define the steps in the genetic evaluation process.
4. List the three procedures that can be done prenatally to diagnosis a fetus with a chromosome abnormality.

LESSON 4 POSTTEST

1. List three real or perceived barriers that might prevent a person from accessing genetic services.
2. List three things you can do to promote the patient-provider interaction.

ANSWERS

LESSON 1 POSTTEST: ANSWERS

1. 45,XX,t(13;21)
2. 46,XY,t(7;10)(p12;q22)
3. 47,XY,+21
4. 45,X
5. 46,XX,inv(10)(p12;q22)
6. True
7. True
8. Blood
9. Skin
10. Product of conception
11. Amniotic fluid
12. Bone marrow

LESSON 2 POSTTEST: ANSWERS

1. Tell your patient why you need to collect family history information and how this information will be used. Obtain permission to ask questions.
2. You should learn their age, sex and health status.
3. Individuals belonging to some ethnic groups are more likely to carry certain genes coding for recessive genetic conditions. When possible, members of these high-risk groups should be offered the option of carrier testing. African Americans, for instance, should be offered the option of carrier testing for the gene coding for sickle cell anemia, Asians may choose to be screened for the genes coding for thalassemia, etc.
4. Knowing that a couple is related is helpful when analyzing a family history. Persons who have a common ancestor are more likely to carry the same recessive genes. Should they choose to have children, their risk of having a child with a recessive disorder is increased.
5. Autosomal recessive; horizontal pattern of inheritance, parents second cousins, both sexes are affected; PKU, sickle cell anemia
6. Autosomal dominant; vertical mode of inheritance, both sexes affected; HD, achondroplasia
7. Chromosomal; multiple miscarriages; translocation 14;21
8. X-linked; males affected, inherited through mothers; hemophilia, color blindness
9. The effects that a teratogenic agent will have on fetal development are determined by: (1) the degree of exposure, (2) the time during gestation the exposure occurred, and (3) the genetic susceptibility of the mother and the fetus.
10. The answers to this question will vary. This list may include questions about the father's

family history; the use of nutritional supplements; hobbies, crafts or potential work exposures; etc.

11. False If a person has a dominant single gene disorder, there is a 50% chance with each pregnancy that he/she will pass on the gene coding for this condition.
12. False There is a 25% chance with each pregnancy that parents who carry the same recessive gene will pass on a copy of this gene and have an affected child.
13. True Growth retardation, developmental delay, and dysmorphic features are characteristics commonly seen in children who have a chromosome abnormality.
14. True A history of three or more pregnancy losses suggests that one of the partners may have a balanced chromosome rearrangement.

LESSON 3 POSTTEST: ANSWERS

1. False Genetic counseling is a nondirective process. Patients are provided with information about all of their reproductive choices and are then responsible for making a decision that best fits their beliefs and values.
2. False Every pregnant woman should be offered the option of MSAFP or triple screening. Whether they pursue testing or not is their choice.
3. Individuals who are referred to a genetics clinic will be asked to provide family history information and sign release of information forms for medical and possibly school records. A physical examination will be done by a clinical geneticist. After a diagnosis is made, time will be spent talking to the patient about the diagnosis, prognosis and available treatment options, if any. They will also be counseled about their occurrence or recurrence risk and the necessary follow-up. If a diagnosis is suspected, laboratory tests may be ordered to confirm the diagnosis. If no diagnosis is made, the patient might be followed on an annual or semiannual basis.
4. The procedures are CVS, early amniocentesis, amniocentesis and ultrasound examination. Maternal serum marker studies are not diagnostic tests. They are screening tests and will only identify women who have an increased risk of having a child with a chromosome abnormality or a structural abnormality that alters the level of maternal serum markers.

LESSON 4 POSTTEST: ANSWERS

1. The answer to this question will vary, however, the list could include (1) financial constraints, (2) distance from the clinic, (3) lack of child care for the unaffected sibs, (4) family or community rules, (5) cultural beliefs, (6) grief, (7) the need to cope with a more immediate crisis, etc.
2. The answer to this question will also vary. Some possible responses include:
   (1) let patients or their parents establish the agenda, or direction of discussion,
   (2) ask open-ended questions, (3) recognize and respond to non-verbal cues,
   (4) assess your personal values and determine whether they might interfere with your ability to work with certain patients, (5) facilitate referral to community programs or support groups, etc.

PENUNTUN BELAJAR (*Learning Guide*)

Lakukan penilaian kinerja pada setiap langkah / tugas dengan menggunakan skala penilaian di bawah ini:		
1	**Perlu perbaikan**	Langkah atau tugas tidak dikerjakan secara benar, atau dalam urutan yang salah (bila diperlukan) atau diabaikan
2	**Cukup**	Langkah atau tugas dikerjakan secara benar, dalam urutan yang benar (bila diperlukan), tetapi belum dikerjakan secara lancar
3	**Baik**	Langkah atau tugas dikerjakan secara efisien dan dikerjakan dalam urutan yang benar (bila diperlukan)

Nama peserta	Tanggal
Nama pasien	No Rekam Medis

PENUNTUN BELAJAR GENETIKA KLINIS							
No.	**Kegiatan / langkah klinik**	**Kesempatan ke**					
		1	**2**	**3**	**4**	**5**	
I.	**Ketrampilan Klinis**						
1.	Gather genetic family history information, including an appropriate multigenerational family history						
2.	Identify clients who would benefit from genetic services						
3.	Explain basic concepts of probability and disease susceptibility, and the influence of genetic factors in maintenance of health and development of disease						
4.	Seek assistance from and refer to appropriate genetics experts and peer support resources						
5.	Obtain credible, current information about genetics, for self, clients, and colleagues.						
6.	Use effectively new information technologies to obtain current information about genetics						
7.	Educate others about client-focused policy issues						
8.	Participate in professional and public education about Genetics						
9	Educate clients about availability of genetic testing and/or treatment for conditions seen frequently in practice						
10.	Provide appropriate information about the potential risks, benefits, and limitations of genetic testing						
11.	Provide clients with an appropriate informed-consent process to facilitate decision making related to genetic testing						
12	Educate clients about the range of emotional effects they and/or family members may experience as a result of receiving genetic information						

13.	Explain potential physical and psychosocial benefits and limitations of gene-based therapeutics for clients					
14.	discuss costs of genetic services, benefits and potential risks of using health insurance for payment of genetic services, potential risks of discrimination					
15.	safeguard privacy and confidentiality of genetic information of clients to the extent possible					
16.	inform clients of potential limitations to maintaining privacy and confidentiality of genetic information					

DAFTAR TILIK

Berikan tanda ✓ dalam kotak yang tersedia bila keterampilan/tugas telah dikerjakan dengan memuaskan, dan berikan tanda ✗ bila tidak dikerjakan dengan memuaskan serta T/D bila tidak dilakukan pengamatan

✓	**Memuaskan**	Langkah/ tugas dikerjakan sesuai dengan prosedur standar atau penuntun
✗	**Tidak memuaskan**	Tidak mampu untuk mengerjakan langkah/ tugas sesuai dengan prosedur standar atau penuntun
T/D	**Tidak diamati**	Langkah, tugas atau ketrampilan tidak dilakukan oleh peserta latih selama penilaian oleh pelatih

Nama peserta didik	Tanggal
Nama pasien	No Rekam Medis

DAFTAR TILIK GENETIKA KLINIS				
No.	**Langkah / kegiatan yang dinilai**	**Hasil penilaian**		
		Memuaskan	Tidak Memuaskan	Tidak diamati
I.	***All health professionals should:***			
1.	recognize philosophical, theological, cultural, and ethical perspectives influencing use of genetic information and services			
2.	appreciate the sensitivity of genetic information and the need for privacy and confidentiality			
3.	recognize the importance of delivering genetic education and counseling fairly, accurately, and without coercion or personal bias			
4.	appreciate the importance of sensitivity in tailoring information and services to clients' culture, knowledge and language level			
5	seek coordination and collaboration with interdisciplinary team of health professionals			
6.	speak out on issues that undermine clients' rights to informed decision making and voluntary action			
7.	recognize the limitations of their own genetics expertise			
8.	demonstrate willingness to update genetics knowledge at frequent intervals			
9.	recognize when personal values and biases with regard to ethical, social, cultural, religious, and ethnic issues may affect or			

	interfere with care provided to clients			
10.	support client-focused policies			

Peserta dinyatakan:	**Tanda tangan pembimbing**
☐ Layak ☐ Tidak layak melakukan prosedur	(Nama jelas)

PRESENTASI

- *Power points*
- Lampiran : skor, dll

Tanda tangan peserta didik

(Nama Jelas)

Kotak komentar