私立臺北醫學院 九十二 學年度第 一 學期期中(末)考試命(試)題紙

系級	科目	授課教師	考試日期	學號	姓名
醫二	**遺傳學**	**葉健全**	93 年 1 月 13 日第 3 節		

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(A)

選擇題：2 分、填空題：3 分、問答題：9 分

1. Three sets of genes control early development in fruit flies except for:
   A. Maternal effect genes
   B. Homeoic genes
   C. Segmentation genes
   D. Pattern formation genes

2. In all organisms, at some point the embryo's own genes become activated and take over from the maternal genes. In humans, this shift first occurs:
   A. Fertilization stages
   B. One- to four-cell stages
   C. Four- to eight-cell stages
   D. Morula stages

3. About 95% of people with a dominantly inherited disorder called achondroplasia have the same mutation in the:
   A. Fibroblast growth factor receptor gene
   B. Hedgehog gene
   C. Integrin gene
   D. Selectin gene

4. All cells contain some genes that promote apoptosis and other genes that prevent it. Some are _____ that chop up chromosome DNA into nucleosome-length pieces.
   A. Proteases
   B. Exonucleases
   C. Endonucleases
   D. Ligase

5. In presumptive males, the second step in sex determination occurs by the seventh week of development, when the _____ gene produces a protein called testis-determining factor (TDF)
   A. Wilms tumor suppressor protein 1 (WT1)
   B. Steroidogenic factor 1 (SF1)
   C. Sex determining region of the Y chromosome (SRY)
   D. Wingless integrated (WNT)

6. Which of following is wrong?
   A. A benign tumor remains localized in the place of origin, often separated from the surrounding cells by a layer of connective tissue
   B. Although compact and slow growing, benign tumors may eventually contain billions of cells. They are invasive, damaging adjacent tissues and often producing internal bleeding
   C. A malignant tumor become disorganized and may revert to a more primitive form, losing their specialized structures and functions
   D. Cancer cells almost always have abnormal karyotypes. These include translocations, inversions, deletions, isochromosomes, monosomies, and extra chromosomes.

7. The *AA1* tumor gene has been cloned, and the AA1 protein product is a transcription factor. The AA1 proteins belong to a class called zinc-finger proteins. The *AA1* gene in their unmutated dominant forms act as brakes on cell division. Mutations of these genes to recessive alleles or their loss promote cancer in somatic cells only when they become homozygous and release the brake. We call these genes:
   A. Tumor suppresser genes
   B. Proto-oncogenes
   C. DNA repair genes
   D. Viral-oncogenes

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8. The X gene encodes normal X protein acts as a major brake on cell division. When phosphate groups are added to X, it releases a transcription factor that eventually turns on genes whose protein products are needed for DNA synthesis. This X gene is
   A. RB gene
   B. WT1 gene
   C. P53 gene
   D. Ras gene

9. The Y proteins referred to earlier are similar to G proteins. In their active form, they pass a signal to additional relay molecules that transverse the cytoplasm. The Y proteins encoded by the oncogenic alleles are stuck in the active form. Thus, they tell the cell to divide inappropriately. This Y gene is
   A. RB gene
   B. WT1 gene
   C. P53 gene
   D. Ras gene

10. The Rous sarcoma virus is one of a family of viruses called
   A. Adenovirus
   B. Parvovirus
   C. Retrovirus
   D. Reovirus

11. In the mid-1980s, geneticists isolated and sequenced the genes and gene products of some segmentation and homeotic loci. They shared a highly similar sequence of about 180 DNA base pairs that was dubbed the _____. These homeodomain-containing proteins are _____ that regulate the turning on or turning off of other genes by binding to their enhancer or promoter regions.

12. Cells bind to each other, move, migrate, divide, differentiate, and die in precisely patterned ways. Research begun in the 1970s has uncovered key proteins known as _____(CAMs)

13. There are two sets of primitive duct systems, the male-type _____ and the female-type _____. These duct systems will eventually form the rest of the internal sex organs.

14. The encoded proteins act in information pathways that extend from the cell surface to nuclear genes—specifically those genes that control growth and division. This line of chemical communication from outside a cell across the plasma membrane, cytoplasm, and nuclear membrane is called _____.

15. Cancer cells may be shed from the primary site and circulate through the blood and lymph to other locations in the body. The spreading to distant sites, called _____.

16. 請說明何謂"oncogene"及"proto-oncogene"，並舉一個實際例子加以說明。

(B)

True and False

1. The thymus gland is important in the development of immune cells and is gradually regress during adolescence.
2. Nonresponsiveness to self antigens is the phenomenon called immunological tolerance.
3. According to Burnet's clonal selection theory, T cells are preprogrammed to recognize and respond to just one antigen.
4. Helper T cell receptor recognizes the antigen on the B cell MHC class I protein, which triggers release of interleukin 2.
5. There are 4 families of closely linked elements: V,D,J, and C in light chain gene K.
6. The plasma cell-cancer cell fusion and the resultant clone is called a hybridoma.
7. In blood group ABO system, the O allele is recessive to A and to B, whereas the A and B alleles are codominant to each other.

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8. Since type C blood is the universal donor, it is safe to use the blood without testing for compatibility by cross matching.
9. Maternal-fetal incompatibility is never a problem for ABO blood group.
10. The lymphocytes in the donor blood marrow mount a pervasive and painful immunological attack against antigens on the cells of the recipient is the *graft-versus-host* disease.
11. Autoimmune diseases have a mix of genetic and environmental causes.
12. The particular alleles of the linked HLA genes are inherited as a group called a haplotype.

(C)

選擇題

(1) 我國罕見疾病之定義, which of following is Incorrect? **(3 分)**
- **A.** 於民國 89 年 8 月 9 日正式施行之「罕見疾病防治及藥物法」有明確之定義
- **B.** 指「疾病盛行率」在行政院衛生署訂定之公告標準以下 (1/20,000)
- **C.** 且經「罕見疾病及藥物審議委員會」認定者
- **D.** 或因情況特殊, 經行政院衛生署指定公告者
- **E.** 審議認定罕見疾病原則:以罕見，遺傳性，診治上困難 爲優先考量

(2) Which one of following is Incorrect about the genetics principles: **(2 分)**
- **A.** Feasibility of prenatal diagnosis
- **B.** Pre-symptomatic testing
- **C.** Population screening
- **D.** Cannot detection of carrier status
- **E.** Gene therapies

(3) Newborn Screening item in Taiwan, which one of following is not including? **(2 分)**
- **A.** G6PD - deficiency
- **B.** Congenital hypothyroidism (CHT)
- **C.** Congenital adrenal hyperplasia(CAH)
- **D.** Homocystinuria (HCU)
- **E.** Galactosemia (GAC)

(4) Matching: (10 分) (請將 a. b. c. d. e. 寫在答案卷上)
- **1.** Cytogenesis:
- **2.** Molecular & biochemical genetics:
- **3.** Genomics:
- **4.** Clinical genetics:
- **5.** Genetic counselign:

- **a.** Study of the structure & function of individual genes.
- **b.** Study of the genome, it's organization & functions.
- **c.** Study of chromosome.
- **d.** Provision of risk information and the care of patients and their families.
- **e.** Application of genetics to diagnosis and patient care..

(5) Matching: (8 分) (請將 a. b. c. d. 寫在答案卷上)

Diseases:
- **1.** Duchenne muscular dystrophy (DMD)
- **2.** Fragile X syndrome
- **3.** Prader-willi syndrome
- **4.** MPS (Mucopolysaccharidoses)

Inheritance pattern:
- **a.** X-Linked (Xq 27.3) and triplet repeat expansion disease
- **b.** Absence of paternally derived 15q 11-13, Uniparental disomy (UPD)
- **c.** Lysosomal storage disorders,11 enzymes defects cause7 types of disorders
- **d.** X-Linked recessive (XLR)

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(A)

1.	2.	3.	4.	5.	6.	7.	8.	9.	10.
11.		12.		13.		14.		15.	

16.

(B)

1.	2.	3.	4.	5.	6.	7.	8.	9.	10.	11.	12.

(C)

1.	2.	3.	4-1.	4-2.	4-3.	4-4.	4-5.	5-1.	5-2.	5-3.	5-4.