PATHOLOGY AIIMS PYQ

Medsynapse by Dr. Nikita

The phenomenon where subsequent generations are at the risk of earlier and more severe disease is known as:

a. Mosaicism
b. Imprinting
c. Pleiotropy
d. Anticipation

Genetic terms

TERM	DEFINITION	EXAMPLE
Codominance	Both alleles contribute to the phenotype of the heterozygote.	Blood groups A, B, AB; α_1-antitrypsin deficiency; HLA groups.
Variable expressivity	Patients with the same genotype have varying phenotypes.	Two patients with neurofibromatosis type 1 (NF1) may have varying disease severity.
Incomplete penetrance	Not all individuals with a disease show the disease. % penetrance × probability of inheriting genotype = risk of expressing phenotype.	*BRCA1* gene mutations do not always result in breast or ovarian cancer.
Pleiotropy	One gene contributes to multiple phenotypic effects.	Untreated phenylketonuria (PKU) manifests with light skin, intellectual disability, musty body odor.
Anticipation	Increased severity or earlier onset of disease in succeeding generations.	Trinucleotide repeat diseases (eg, Huntington disease).
Loss of heterozygosity	If a patient inherits or develops a mutation in a tumor suppressor gene, the wild type allele must be deleted/mutated/eliminated before cancer develops. This is not true of oncogenes.	Retinoblastoma and the "two-hit hypothesis," Lynch syndrome (HNPCC), Li-Fraumeni syndrome.
Epistasis	The allele of one gene affects the phenotypic expression of alleles in another gene.	Albinism, alopecia.
Aneuploidy	An abnormal number of chromosomes; due to chromosomal nondisjunction during mitosis or meiosis.	Down syndrome, Turner syndrome, oncogenesis.

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A 10-year old boy was presented with a mass in abdomen. On imaging, the para-aortic lymph nodes were enlarged. On biopsy, starry sky appearance was seen. What is the underlying abnormality?

a) p53 gene mutation
b) Rb gene mutation
c) Translocation involving BCR-ABL gene
d) Translocation involving Myc gene

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After an incised wound, new collagen fibrils are seen along with a thick layer of growing epithelium. The approximate age of the wound is:

a) 4–5 days
b) About 1 week
c) 12–24 hours
d) 24–72 hours

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PHASE OF WOUND HEALING	EFFECTOR CELLS	CHARACTERISTICS
Inflammatory (up to 3 days after wound)	Platelets, neutrophils, macrophages	Clot formation, ↑ vessel permeability and neutrophil migration into tissue; macrophages clear debris 2 days later
Proliferative (day 3–weeks after wound)	Fibroblasts, myofibroblasts, endothelial cells, keratinocytes, macrophages	Deposition of granulation tissue and type III collagen, angiogenesis, epithelial cell proliferation, dissolution of clot, and wound contraction (mediated by myofibroblasts) Delayed second phase of wound healing in vitamin C and copper deficiency
Remodeling (1 week–6+ months after wound)	Fibroblasts	Type III collagen replaced by type I collagen, ↑ tensile strength of tissue Collagenases (require zinc to function) break down type III collagen Zinc deficiency → delayed wound healing

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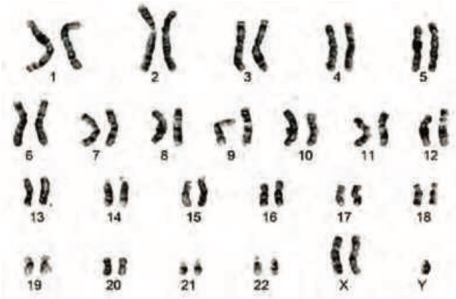
Which of the following abnormality is seen in the given karyotype?



a) High pitched cry
b) Round face with protruding tongue
c) Short stature with webbed neck
d) Gynecomastia with long thin limbs

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Klinefelter syndrome

Male, 47,XXY.
Small, firm testes; infertility (azoospermia); tall stature with eunuchoid proportions (delayed epiphyseal closure → ↑ long bone length); gynecomastia; female hair distribution.
May present with developmental delay.
Presence of inactivated X chromosome (Barr body). Common cause of hypogonadism seen in infertility workup. ↑ risk of breast cancer.

Dysgenesis of seminiferous tubules → ↓ inhibin B → ↑ FSH.
Abnormal Leydig cell function → ↓ testosterone → ↑ LH.

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Inheritance of ABO blood group is:

a) X-linked inheritance
b) Autosomal Recessive inheritance
c) Mitochondrial inheritance
d) Codominance

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Most common nephropathy associated with malignancy is:

a. Focal segmental glomerulosclerosis (FSGS)
b. Minimal change disease
c. IgA nephropathy
d. Membranous glomerulonephritis

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Membranous nephropathy	Also called membranous glomerulonephritis. Can be 1° (eg, antibodies to phospholipase A_2 receptor) or 2° to drugs (eg, NSAIDs, penicillamine, gold), infections (eg, HBV, HCV, syphilis), SLE, or solid tumors. ↑ risk of thromboembolism (eg, DVT, renal vein thrombosis).	Diffuse capillary and GBM thickening C	Granular due to immune complex (IC) deposition	"Spike and dome" appearance of subepithelial deposits

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Oil red 'O' stain is used for:

a) Glutaraldehyde fixed specimen
b) Alcohol fixed specimen
c) Formalin fixed specimen
d) Frozen specimen

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Which of the following is responsible for adhesion of platelets to the vessel wall?

a) Factor IX
b) Von Willebrand factor
c) Fibrinogen
d) Fibronectin

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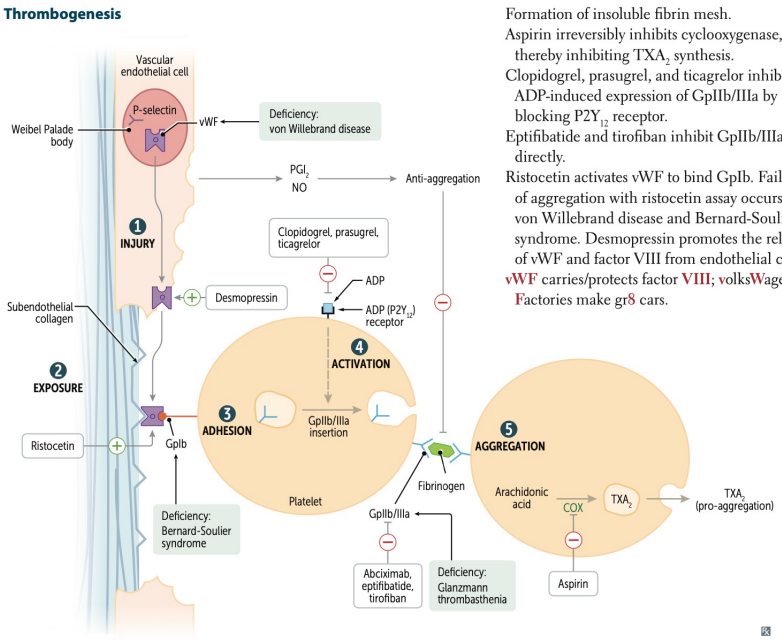

Formation of insoluble fibrin mesh.
Aspirin irreversibly inhibits cyclooxygenase, thereby inhibiting TXA_2 synthesis.
Clopidogrel, prasugrel, and ticagrelor inhibit ADP-induced expression of GpIIb/IIIa by blocking $P2Y_{12}$ receptor.
Eptifibatide and tirofiban inhibit GpIIb/IIIa directly.
Ristocetin activates vWF to bind GpIb. Failure of aggregation with ristocetin assay occurs in von Willebrand disease and Bernard-Soulier syndrome. Desmopressin promotes the release of vWF and factor VIII from endothelial cells.
vWF carries/protects factor VIII; volksWagen Factories make gr8 cars.

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Lymphatic spread is most commonly seen in which type of thyroid malignancy:

a) Papillary carcinoma
b) Follicular carcinoma
c) Medullary carcinoma
d) Anaplastic carcinoma

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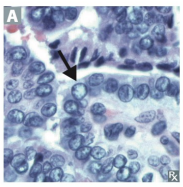
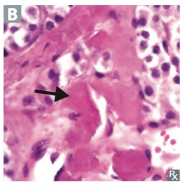
Papillary carcinoma 	Most common. Empty-appearing nuclei with central clearing ("Orphan Annie" eyes) A, psamMoma bodies, nuclear grooves (Papi and Moma adopted Orphan Annie). ↑ risk with *RET/PTC* rearrangements and *BRAF* mutations, childhood irradiation. Papillary carcinoma: most prevalent, palpable lymph nodes. Good prognosis.
Follicular carcinoma	Good prognosis. Invades thyroid capsule and vasculature (unlike follicular adenoma), uniform follicles; hematogenous spread is common. Associated with *RAS* mutation and *PAX8-PPAR-γ* translocations. Fine needle aspiration cytology may not be able to distinguish between follicular adenoma and carcinoma.
Medullary carcinoma 	From parafollicular "C cells"; produces calcitonin, sheets of polygonal cells in an amyloid stroma B (stains with Congo red). Associated with MEN 2A and 2B (*RET* mutations).
Undifferentiated/ anaplastic carcinoma	Older patients; presents with rapidly enlarging neck mass → compressive symptoms (eg, dyspnea, dysphagia, hoarseness); very poor prognosis. Associated with *TP53* mutation.

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This image was taken by attaching the camera to the microscope. What is the requirement for such a microscope?

a) Dark field condenser
b) Phase shifter
c) Dichroic mirror
d) Cathode ray tube

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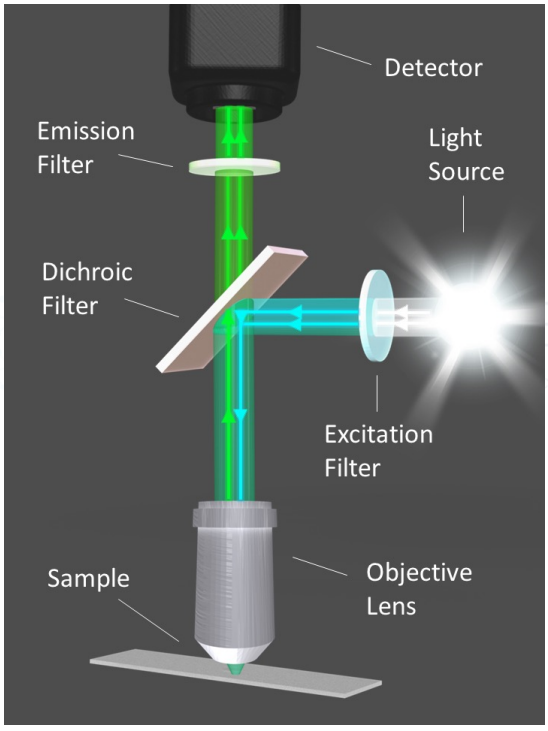

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In Langerhans Cell Histiocytosis, the characteristic abnormality seen is:

a) Foamy macrophages
b) Giant cell
c) Plasma cell
d) Birbeck's granules

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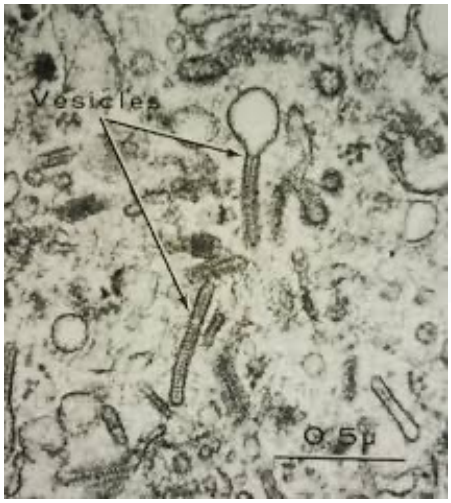

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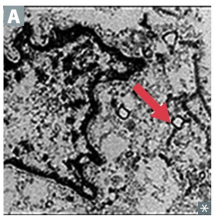
Langerhans cell histiocytosis



Collective group of proliferative disorders of Langerhans cells (antigen-presenting cells normally found in the skin). Presents in a child as lytic bone lesions and skin rash or as recurrent otitis media with a mass involving the mastoid bone. Cells are functionally immature and do not effectively stimulate primary T cells via antigen presentation. Cells express S-100 and CD1a. Birbeck granules ("tennis rackets" or rod shaped on EM) are characteristic A.

Type of multifocal, unisystem Langerhans cell histiocytosis, as demonstrated by HPE.

Triad of:

- Lytic bone lesions (often in skull)
- Exophthalmos
- Diabetes insipidus (due to pituitary stalk involvement)

[Seen in only one-third of patients]