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AR: Bloom FAX Rest are AD Investigations • 135

7.2 Inherited cancer predisposition syndromes		Exam night topic	
Syndrome	Malignancies	Inheritance	Gene
Ataxia telangiectasia	Leukaemia, lymphoma, ovarian, gastric, brain, colon	AR	ATM
Bloom syndrome	Leukaemia, tongue, oesophageal, colonic, Wilms' tumour	AR	BLM
Breast/ovarian	Breast, ovarian, colonic, prostatic, pancreatic	AD	BRCA1, BRCA2
Cowden syndrome	Breast, thyroid, gastrointestinal tract, pancreatic	AD	PTEN
Familial adenomatous polyposis	Colonic, upper gastrointestinal tract	AD	APC, MUTYH
Familial atypical multiple mole melanoma (FAMMM)	Melanoma, pancreas	AD	CDKN2A (TP53)
Fanconi anaemia	Leukaemia, oesophageal, skin, hepatoma	AR	FANCA, FANCB, FANCD1
Gorlin syndrome	Basal cell skin, brain	AD	PTCH1
Hereditary diffuse gastric cancer	Diffuse gastric cancer	AD	E-cadherin
Hereditary non-polyposis colon cancer (HNPPCC)	Colonic, endometrial, ovarian, pancreatic, gastro	AD	MSH2, MLH1, MSH6, PMS1, PMS2
Li-Fraumeni syndrome	Sarcoma, breast, osteosarcoma, leukaemia, glioma, adrenocortical	AD	TP53
Multiple endocrine neoplasia (MEN) 1	Pancreatic islet cell, pituitary adenoma, parathyroid adenoma and hyperplasia	AD	MEN1
MEN 2	Medullary thyroid, pheochromocytoma, parathyroid hyperplasia	AD	RET
Neurofibromatosis 1	Neurofibrosarcoma, pheochromocytoma, optic glioma	AD	NF1
Neurofibromatosis 2	Vestibular schwannoma	AD	NF2
Papillary renal cell cancer syndrome	Renal cell cancer	AD	MET
Peutz-Jegher syndrome	Colonic, ileal, breast, ovarian	AD	STK11
Prostate cancer	Prostate	AD	APC1
Retinoblastoma	Retinoblastoma, osteosarcoma	AD	RB1
von Hippel-Lindau syndrome	Haemangioblastoma of retina and CNS, renal cell, pheochromocytoma	AD	VHL
Wilms' tumour	Nephroblastoma, neuroblastoma, hepatoblastoma, rhabdomyosarcoma	AD	WT1
Xeroderma pigmentosum	Skin, leukaemia, melanoma	AR	XPA, XPC, XPB, ERCC1, XPD

(AD = autosomal dominant, AR = autosomal recessive, CNS = central nervous system)

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ARDMS Abdomen Sonography Examination Sample Questions (Q154-Q159):

NEW QUESTION # 154

Which change of the inferior vena cava spectral Doppler waveform is expected superior to a nonocclusive thrombus?

- **A. Dampening**
- B. Absence of flow
- C. Increased velocity
- D. Becomes multiphasic

Answer: A

Explanation:

In the presence of a nonocclusive thrombus, Doppler waveform above the thrombus typically shows dampened flow with loss of normal respiratory phasicity due to partial venous outflow obstruction. Complete absence of flow is typically seen with occlusive thrombus.

According to Zwiebel's Introduction to Vascular Ultrasound:

"Partial obstruction produces dampened and continuous flow patterns superior to a nonocclusive thrombus." Reference:

Zwiebel WJ, Pellerito JS. Introduction to Vascular Ultrasound. 6th ed. Elsevier, 2019.

AIUM Practice Parameter for Venous Ultrasound, 2020.

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NEW QUESTION # 155

Which vascular condition is most commonly associated with a wandering spleen?

- **A. Torsion**
- B. Infarction
- C. Rupture
- D. Portal hypertension

Answer: A

Explanation:

A wandering spleen occurs when the spleen is not adequately anchored by its supporting ligaments, allowing it to move freely within the abdomen. This increases the risk of splenic torsion, which compromises vascular supply and may result in infarction if not corrected.

According to Rumack's Diagnostic Ultrasound:

"The most serious complication of a wandering spleen is torsion, which may result in splenic infarction." Reference:

Rumack CM, Wilson SR, Charboneau JW, Levine D. Diagnostic Ultrasound. 5th ed. Elsevier, 2017.

Moore KL, Clinically Oriented Anatomy. 8th ed. Wolters Kluwer, 2018.

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NEW QUESTION # 156

Which renal condition is commonly associated with pyuria and leukocytosis?

- A. Staghorn calculus
- B. Nephrocalcinosis
- C. Renal cell carcinoma
- **D. Acute pyelonephritis**

Answer: D

Explanation:

Acute pyelonephritis is a bacterial infection of the renal parenchyma and collecting system. Classic clinical findings include fever, flank pain, leukocytosis (elevated white blood cells), and pyuria (white blood cells in urine). Ultrasound may demonstrate renal enlargement, decreased echogenicity, and loss of corticomedullary differentiation.

* Nephrocalcinosis (A) involves calcium deposition without infection.

* Staghorn calculus (B) may lead to infection but is primarily characterized by obstructive uropathy.

* Renal cell carcinoma (C) presents with hematuria and mass formation rather than infection symptoms.

Reference Extracts:

* Rumack CM, Wilson SR, Charboneau JW, Levine D. Diagnostic Ultrasound. 5th ed. Elsevier, 2017.

Chapter: Kidneys.

* Middleton WD, Kurtz AB, Hertzberg BS. Ultrasound: The Requisites. 3rd ed. Elsevier, 2015.

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NEW QUESTION # 157

Which of the following is a possible early complication of a renal transplant?

- A. Transitional cell carcinoma
- B. Acute tubular necrosis
- C. Transplant artery stenosis
- D. Ureterocele

Answer: B

Explanation:

Acute tubular necrosis (ATN) is the most common cause of early graft dysfunction following renal transplantation. It results from ischemia-reperfusion injury during the transplantation process. Ultrasound findings may be nonspecific but Doppler may show elevated resistive indices.

Ureterocele (A) is a congenital anomaly.

Transplant artery stenosis (C) is a late complication.

Transitional cell carcinoma (D) is rare and not typically an early complication.

Reference Extracts:

Middleton WD, Kurtz AB, Hertzberg BS. Ultrasound: The Requisites. 3rd ed. Elsevier, 2015.

Rumack CM, Wilson SR, Charboneau JW, Levine D. Diagnostic Ultrasound. 5th ed. Elsevier, 2017.

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NEW QUESTION # 158

Which of the following must be sterile for a percutaneous procedure?

- A. Transducer
- B. Transducer cover
- C. Gel within transducer cover
- D. Machine controls

Answer: C

Explanation:

In percutaneous procedures such as biopsies or drainages, maintaining a sterile field is critical to avoid introducing infection. While the transducer is covered by a sterile cover, the gel placed inside this cover (between the probe and the cover) must also be sterile, as it contacts the sterile field. The transducer itself and machine controls are not sterile but are handled appropriately to avoid field contamination.

According to the AIUM Guidelines:

"Sterile coupling gel must be used inside the sterile probe cover during all invasive or percutaneous procedures." (AIUM Guidelines for Cleaning and Preparing Ultrasound Transducers, 2021).

Reference:

AIUM Guidelines for Cleaning and Preparing Ultrasound Transducers and Equipment for Reuse, 2021.

ACR Practice Parameter for Performing Ultrasound-Guided Procedures, 2020.

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NEW QUESTION # 159

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