

Quiz

ERN-Skin Session at EADV Congress



ABOUT XERODERMA PIGMENTOSUM

1. Which cancers have been associated with xeroderma pigmentosum type C (XP)?

Select all that apply.

- A. Basal cell carcinoma
- B. Squamous cell carcinoma
- C. Melanoma
- D. Certain internal malignancies
- E. Thyroid cancer exclusively

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ABOUT XERODERMA PIGMENTOSUM

2. Which statements regarding internal malignancies in XP type C are correct?

Select all that apply.

- A. Internal malignancies have been reported in XP
- B. The risk of internal malignancies is generally less prominent than the risk of skin cancer
- C. The underlying DNA repair defect provides a biological basis for cancer susceptibility
- D. XP predisposes exclusively to cutaneous cancers.
- E. Internal malignancies can occur independently of direct UV exposure

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ABOUT XERODERMA PIGMENTOSUM

3. Which types of internal malignancies have been reported in patients with XP-C?

Select all that apply.

- A. Central nervous system tumors
- B. Hematological malignancies
- C. Gastrointestinal malignancies
- D. Certain solid tumors outside the skin
- E. No internal malignancies have ever been reported

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ABOUT NEUROFIBROMATOSIS TYPE 1 (NF1)

4. An 18-year-old patient with neurofibromatosis type 1 (NF1) and a known plexiform neurofibroma of the thigh reports new persistent pain and noticeable enlargement of the lesion over the past few weeks. On examination, the lesion is firm on palpation. What is the most appropriate next step?

- A. Reassure the patient and reassess the lesion at the next routine annual visit.
- B. Promptly investigate the lesion for possible malignant transformation, including regional MRI and appropriate metabolic imaging (e.g. FDG-PET-MRI), and refer to an experienced multidisciplinary NF1 team.
- C. Initiate treatment with a MEK inhibitor without further diagnostic assessment.
- D. Focus treatment on the patient's cutaneous neurofibromas, as these represent the principal tumour-related concern during transition to adulthood.

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~~A. Reassure the patient and reassess the lesion at the next routine annual visit.~~

Incorrect: Rapid growth, new or persistent pain, particularly nocturnal or difficult-to-control pain, and a change in tumour consistency are warning signs for atypical neurofibromatous neoplasms or malignant peripheral nerve sheath tumour (MPNST) and should not be managed by routine observation alone.

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B. Promptly investigate the lesion for possible malignant transformation, including regional MRI and appropriate metabolic imaging (e.g. FDG-PET-MRI), and refer to an experienced multidisciplinary NF1 team.

Correct: New pain, rapid enlargement and increasing firmness of a plexiform neurofibroma should prompt investigation for malignant transformation. Regional MRI is the initial imaging modality, with FDG PET-MRI or PET-CT used as appropriate when malignancy is suspected. Further diagnostic and therapeutic decisions should be made within an experienced multidisciplinary NF1 team.

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~~C. Initiate treatment with a MEK inhibitor without further diagnostic assessment.~~

Incorrect: MEK inhibition can be considered for selected symptomatic, inoperable plexiform neurofibromas. However, suspected malignant transformation requires diagnostic evaluation before treatment of the lesion as a benign plexiform neurofibroma.

~~D. Focus treatment on the patient's cutaneous neurofibromas, as these represent the principal tumour-related concern during transition to adulthood.~~

Incorrect: Cutaneous neurofibromas may cause considerable cosmetic and psychosocial burden and should be addressed during transition care. However, a changing, painful plexiform neurofibroma takes clinical priority because of the possibility of malignant transformation.”

ABOUT DARIER DISEASE

5. Which mucosal membrane is not involved in Darier disease:

Select all that apply.

- A. Oral cavity: Asymptomatic papules with a cobblestone appearance involving the palate, tongue, or gingiva
- B. Ocular: Conjunctival hyperaemia, blepharitis, or marginal lesions of the eyelids
- C. Genital: Keratotic pink to red–brown papules or plaques, often pruritic and occasionally associated with dyspareunia
- D. Esophagus: Inflammation leading to dysphagia

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ABOUT DARIER DISEASE

6. Which biologics have been more effective in Darier disease, although with variable results:

Select all that apply.

- A. Anti-IL17
- B. Anti IL4/13
- C. Anti IL-13
- D. Anti-IL1

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ABOUT GENETICS

7. Which finding most strongly supports hepatoerythropoietic porphyria (HEP) rather than typical porphyria cutanea tarda?

Select all that apply.

- A. Acute abdominal pain with neuropsychiatric symptoms
- B. Severe photosensitivity beginning in childhood with biallelic pathogenic UROD variants
- C. Painful, non-blistering photosensitivity beginning in adulthood
- D. Blistering confined to sun-exposed skin with normal urinary porphyrins

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B. Severe photosensitivity beginning in childhood with biallelic pathogenic UROD variants

Correct: HEP results from severe UROD deficiency caused by biallelic pathogenic variants. It typically begins in childhood and causes pronounced photosensitivity, skin fragility, blistering, scarring and hypertrichosis.

~~C. Painful, non-blistering photosensitivity beginning in adulthood~~

~~D. Blistering confined to sun-exposed skin with normal urinary porphyrins~~

ABOUT DARIER DISEASE

8. After failure of corticosteroids with dapsone, cyclosporine or MMF, which treatment was effective and well tolerated in this patient?

Select all that apply.

- A. Escalation of systemic corticosteroids
- B. Methotrexate monotherapy
- C. IVIG as adjunctive therapy
- D. Higher-dose cyclosporine

ABOUT DARIER DISEASE

8. After failure of corticosteroids with dapsone, cyclosporine or MMF, which treatment was effective and well tolerated in this patient?

~~Select all that apply.~~

~~A. Escalation of systemic corticosteroids~~

Incorrect: Prior corticosteroid-based regimens failed and caused adverse effects.

~~B. Methotrexate monotherapy~~

Incorrect: Not used in this patient; the case provides no evidence of benefit.

C. IVIG as adjunctive therapy

Correct: 400 mg/kg/day for 5 days: effective and well tolerated as an adjunct.

~~D. Higher-dose cyclosporine~~

Incorrect: Cyclosporine had already failed; escalation could increase toxicity.

ABOUT MOSAICISM

9. What is the main feature that allows the differential diagnosis between CMTC and Sturge-Weber syndrome?

Select all that apply.

- A. The anatomical site involved
- B. The presence or absence of a reticular pattern
- C. The presence or absence of cutaneous atrophy
- D. The presence or absence of limb hypotrophy

ABOUT MOSAICISM

9. What is the main feature that allows the differential diagnosis between CMTC and Sturge-Weber syndrome?

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C. The presence or absence of cutaneous atrophy

Correct: The correct answer is the presence or absence of cutaneous atrophy, as skin atrophy is typically associated with CMTC. In contrast, in Sturge-Weber syndrome associated with GNA11 variants, capillary malformations with a reticular pattern, limb hypotrophy, and involvement of different anatomical sites may also occur, as in CMTC. Therefore, these features are less useful for differential diagnosis

~~D. The presence or absence of limb hypotrophy~~

**THE WINNER
IS...**



**Alessandra
Gelmetti**

