

DNA Subthematic Group

PATIENT PATHWAYS

When a diagnosis of Xeroderma Pigmentosum should be suggested ?

- Lentigines, modification of the skin
- Sun sensitivity
- Early skin Cancer

Refer the patient to an Expert Health Care

Confirmation of the diagnosis

Genetic testings

Multigene Panel Testing

MULTIGENE PANEL TESTING INCLUDING THE GENES *DB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA and XPC*

Evaluation of a patient at diagnosis

SKIN

- Perform baseline examination of the skin
- Examination of the scalp
- Examination of the lip and adjacent tip of the tongue for signs of sun damage
- Baseline clinical color photographs of the entire skin surface

NON SKIN

- Ophthalmologic evaluation
- Neurological evaluation (Deep tendon reflex testing, Measurement of the occipital frontal circumference (OFC), MRI of the brain if other neurologic problems are detected)
- Auditory Baseline audiometry
- Genetic. Consultation with a clinical geneticist and/or genetic counselor

Follow-up

SKIN

Examination of the skin at frequent intervals (every ~3-6 months, depending on the severity of skin disease).

Education of the patients for early recognition of the lesions : abnormal pigmented lesion, appearance a BCC

NON SKIN

- Ophthalmologic evaluation every 6 months
- Neurological evaluation (depending on the type)
- Hearing assessment
- Vitamine D supplementation

EVERYDAY LIFE

DO AND DON'T

DO

Everyday Life organisation

Protection from UV

Mask, clothes

Dosimeter

UV meters are readily available to enable monitoring of areas to identify unexpected UV sources.

DON'T

Artificial sources of UV. (mercury arc, halogen, and other lamps)

SMOKE

