

Painful photosensitivity on sun-exposed skin
with erythema, œdema, petechiae +
stinging, prickling and burning
without blisters



Plasma fluorescence peak at
634 nm testing

If positive



Plasma / erythrocytic
free protoporphyrin concentration +
Faecal protoporphyrin concentration

If increased



Diagnosis: Erythropoietic Protoporphyria

*Or X Linked Dominant Protoporphyria
(If Zn-protoporphyrin % increased ++)*



(Determination of FECH activity level)
if available



Liver examination
Liver function tests, viral
hepatitis serology,
hemochromatosis test,
serum markers for hepatic
fibrosis, Liver imaging
Urinary coproporphyrin

FECH mutation + IVS3-48C
allele determination
ALAS2 mutations

Genetic study of the family