Genus database

21075 poikiloderma sclerosing hereditary

Eponyms: supposes autosomal dominant

Inheritance: supposes autosomal dominant

Semeiological Synthesis: Cutaneous disorder, isolated defect. Palms/soles sclerosis, hyperkeratosis, finger clubbing, generalized poikiloderma characterized by hypo/hyperpigmentation, with telangectasia and atrophy, subcutaneous calcification.

Group Sub group Signs:  
DERMATOLOGICAL DISORDERS  
cuts, changes in appearance and/or features  
- isolated dermatological disorders  
cuts, dysplastic, not including ectodermal dysplasia  
- cuts, scarring  
keratinisation defects  
- keratosis, hyperkeratosis, keratoderma, scaling cuts  
pigmentation changes  
- cuts, speckled, patchy, pitted, marbled  
- poikiloderma

OROCRANIOFACIAL ANOMALIES  
cuts, vascular changes  
- angiectasia, telangiectases

SKELETAL DISORDERS  
fingers, modified form, deformity  
- fingers, broad, clubbing, including spatulate, stubbing, bullous  
ossification, changes  
- calcification ectopic not including: intracranial calcifications (calcinosis)

Super group: 
Super agggreg. Aggregations: OTHER  
isolated dermatological disorders

Differential diagnosis:  
4060 calcinosi tumoural  
13970 Kindler disease  
22920 Rothmund-Thomson syndrome  
27060 xeroderma pigmentosum I

Bibliography OMIM ID: 173700  