

A new ERA for global Dermatology 10 - 15 JUNE 2019 MILAN, ITALY

GENETICS AND GENODERMATOSES

PSEUDOXANTHOMA ELASTICUM- IMPACT ON WORKING CONDITIONS AND QUALITY OF LIFE

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Pseudoxanthoma elasticum (PXE) is a rare inherited multisystem disease (OMIM #177850), caused by mutations in the ABCC6 gene. Various symptoms in skin, eye and cardiovascular system have been previously described, which include yellowish cobblestone papules and elastosis cutis in axillary folds, neck, enoral lower lip or genital mucosa. Diagnosis is dermatologically conclusive and for decades has been confirmed histopathologically; molecular testing is current standard of care. Cardiovascular involvement includes early onset atherosclerosis accompanied by increased rates of vascular occlusions. Treatment consists of walking exercise, medicinal approaches and interventional revascularisation. Ocular involvement includes choroidal neovascularization often treated with anti-vascular endothelial growth factor (anti-VEGF), but treatment decisions remain challenging. For diagnostic accuracy and follow-up of neovascular activity, optical coherence tomography (OCT), fluorescein angiography and OCT angiography (OCT-A) are potent imaging modalities. Due to little prevalence, data on quality of life are rare. We recruited 26 patients with support of "PXE Selbsthilfegruppe Deutschland". Quality of life (DLQI with formal permission) in PXE patients wasn't affected in 16%, 28% suffered small effects, 32% moderate, 22% very large and 4% extremely large effects. 28% of patients reported impairment on work issues, 20% minor effects, 20% no effects. 24 patients weren't able to consult a occupational physician concerning PXE. Disability reintegration management was used in two cases, one of which wasn't PXE-related. Working hazard assessment was evaluated in no case. Invalidity pension was granted in 2 cases. Severe disability was officially acknowledged in 52% (13 patients) ranging from 30-100%. Five patients used medical aid like blue light-filtering. Three patients used tele-workplace fully





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and two partially. Involvement of occupational physicians and medical aid at early stage is desirable. Homeoffice-workplace to optimize working conditions should be implemented whenever possible. More interdisciplinary engagement is crucial to meet patients' needs.



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