2022-2023

THÈSE

pour le

DIPLÔME D'ÉTAT DE DOCTEUR EN MÉDECINE

ET SANTÉ AU TRAVAIL

Employment of Patients with Pseudoxanthoma Elasticum (PXE). A survey in the French reference center for PXE patients

Problématiques de maintien en emploi chez les patients atteints de Pseudoxanthome Élastique

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Soutenue publiquement le :
23 Mars 2023

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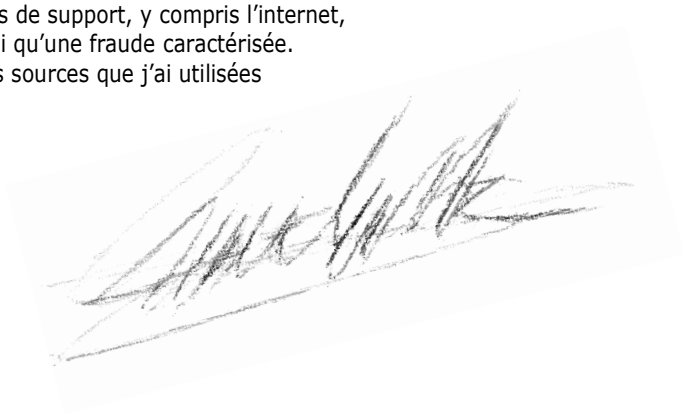

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REMERCIEMENTS

Charlotte LEUGER née REGNAULT :

Merci à tous ceux que je n'ai pu citer. Par ordre antéchronologique :

Merci au **Dr Stevenard,** au **Dr Boissel** pour votre compréhension et la liberté que vous m'avez donnée pour travailler à ma thèse tout en me formant à notre métier et à **Clémence, Valérie, Céline, Mélanie** et **Caroline** pour le plaisir de ces six mois à travailler avec vous. De façon plus personnelle, **Dr Stevenard**, merci de m'avoir transmis votre dynamique et votre vision du travail en équipe ; **Dr Boissel**, merci de m'avoir transmis votre approche pour notre spécialité et votre précision pour notre travail.

Merci à **Thomas**, le meilleur co-thésard, d'avoir accepté de souffrir avec moi dans ce parcours du combattant. On en aura envoyé des mails, passé des appels et bu des bières ! Merci pour ton écoute, nos échanges de points de vue et d'expériences, les repas, les bons moments et de m'avoir permis de refaire du JDR en étant un aussi bon MJ.

Merci à mon fils, **Henry**, qui par sa nature m'a donné des joies sans commune mesure.

Merci à **Clara**, mon amie qui m'a montré une première thèse et qui a mis la barre très haute ! Merci pour toutes nos heures de discussions bienveillantes et heureuses.

Merci à **Elsa**, qui a su me remonter le moral, m'écouter et m'indiquer des chemins à suivre.

Merci au **Pr Martin** de m'avoir fait confiance il y a 3 ans et de m'avoir donné ce magnifique sujet de thèse. Merci pour votre encadrement et les perspectives ouvertes. J'espère que notre étude ouvrira des actions pérennes et utiles.

Merci à tous mes co-internes pour avoir embelli encore le meilleur internat de France avec une attention spéciale pour **Paul** tes conseils toujours pertinents et justes, **Eric** mon partenaire des ennuis, **Gabrielle** et **Antoine** les premiers de la meilleure promotion et enfin **Geoffrey** et **Pierre** pour les soirées pleines de discussions riches.

Merci au **Pr Roquelaure** pour les discussions passionnantes et pleines d'apprentissage lors de notre passage en stage.

Merci au **Pr Petit** pour tant de choses tant professionnelles que personnelles. Merci pour ton aide, ta patience, ton écoute, ta bonne humeur et j'en passe... Nous te devons beaucoup avec Laurent.

Merci à mon mari, **Laurent**, sans toi je n'existe pas mon ange.

Merci à **Elise** mon amie depuis le premier jour en médecine ! Nous sommes passées ensemble par les pires stages d'externat. Tu m'as fait l'honneur d'être ma témoin de mariage. Que notre amitié soit aussi longue que nos vies.

Merci à **Maryse** et **Pierre**, qui m'ont offert mon premier stéthoscope. Merci pour votre soutien, votre bonne humeur et votre optimisme.

Merci à **Ariane**, un médecin et un être humain exceptionnel, qui m'a montré la voie de la médecine et m'a donné des conseils éclairés qui, j'en suis sûre, m'ont grandement aidé pour la réussite de la PACES.

Merci à mes amis du lycée, **Anaëlle**, **Allan**, **Franck-Alain**, qui sont un soutien heureux et une constante dans ma vie !

Merci à Mme **Zylka**, mon enseignante de primaire, que je n'oublierai jamais et qui a changé tout mon parcours de vie.

REMERCIEMENTS

Charlotte LEUGER née REGNAULT :

A **Nawël** et **Sarah**, je ne me rappelle pas ma vie sans vous mes « sœurs ». Une pensée pour toi courageuse et bienveillante **Zohra**, j'espère que tu auras compris que tu es de notre famille.

A tous mes oncles, tantes, cousines et cousins, ma Famille, que j'aime et dont je suis fière.

A ma grand-mère, **Elisabeth**, ce jour de thèse est aussi pour toi et papy. Votre regard m'a donné du courage pour toujours continuer et j'espère que tu ressentiras autant de fierté que j'en ai à t'appeler « mamy ».

A mon frère, **Tanguy**, que j'aime et qui est toujours présent même quand il n'était pas imaginable qu'il soit présent. Je te souhaite tout le bonheur possible.

Le dernier mot pour mes parents **Nadia** et **Olivier** : par votre amour inébranlable et inconditionnel, vous m'avez permis d'exister, de vivre, de ressentir, d'expérimenter, de me découvrir, de prendre confiance en moi… La vie n'est pas "un long fleuve tranquille" mais il est rempli de votre présence. Votre fille à jamais.

REMERCIEMENTS

Thomas DAVID :

L'un de mes amis m'avait mis en garde : « Prends le temps d'écrire tes remerciements ! ». C'est en débutant l'exercice que l'on se rend effectivement compte de sa difficulté. Comment résumer et remercier, en quelques lignes seulement, les gens qui nous ont aidé à parvenir au terme de 10 années d'études (et 12 dans mon cas). Aussi, j'assurerai mes arrières et commencerai par remercier tous ceux que ma mémoire défaillante oubliera dans ce qui suit.

Au commencement des éloges, je voudrais remercier chaleureusement les professeurs **Audrey PETIT**, **Ludovic MARTIN** et **Yves ROQUELAURE** pour m'avoir permis de toucher d'une petite phalange timide le monde de la recherche en médecine. Pour leurs conseils avisés lors de la rédaction de cet article ; pour nos échanges passionnants lors de mon passage en pathologies professionnelles et pour le gout de la médecine du travail et de la recherche en général qu'ils inspirent à leurs internes, sincèrement, merci.

Mes remerciements suivants s'adressent aux chefs et aux équipes qui m'ont encadré et accompagné lors de mes stages d'internat : je citerai de nouveau les **Pr PETIT** et **ROQUELAURE** ainsi que toute l'équipe de la pathologie professionnelle ; mes cheffes du SMIA les **Dr RABJEAU**, **GRELLIER** et **LOURTIS** qui m'ont permis de confirmer mon choix de spécialité par l'exercice de la santé au travail dans sa réalité de terrain ainsi que les équipes de l'antenne de St Barthelemy. Le **Dr VANNIER Laure** et toute l'équipe du secteur de Loire E, merci de m'avoir fait découvrir la spécialité ô combien difficile mais passionnante de la psychiatrie et de m'avoir accompagné dans un stage difficile sur le plan personnel. Enfin, un remerciement spécial au **Dr BRAYER Arnaud** et à toute l'équipe de TOURNESOL, merci d'avoir été une équipe accueillante, compréhensive et humaine et d'avoir redonné confiance à l'interne apeuré et dépressif que j'étais lors de mon premier semestre, cette thèse est en grande partie la vôtre. Merci.

Terminons le tour du personnel médical en remerciant bien évidement mes co-internes, en commençant tout particulièrement par les anciens internes mais presque docteurs **DENIS Axel** et **LACOUR Emma** pour leurs conseils avisés et leurs avis éclairés, il viendra le jour où nous en aurons fini avec ces études commencées maintenant il y a bien trop longtemps ! Et comment ne pas remercier mes co-internes de Médecine du travail, avec la mention spéciale à la meilleure promotion (et de loin) en les personnes de **NICOLLEAU Pierre** et **LAPORTE Geoffrey**, merci pour la solidarité qui nous a rapidement animés tous les 3, pour le support émotionnel et la tradition des mêmes ; bientôt viendra votre heure donc immense courage !

Enfin, je ne pouvais pas écrire de remerciements sans mentionner la personne qui m'a permis de participer à ce travail incroyable et sans qui, rien de tout ça n'aurait été possible. A ma chère co-interne et amie, **REGNAULT-LEUGER Charlotte**. Merci pour m'avoir proposé de t'aider dans ce travail ; merci pour le soutien psychologique lors de la rédaction de ce papier (et ces appels interminables) ; merci pour les échanges autour des sagas mp3 ou les parties de D&D ; sincèrement merci.

En s'éloignant du spectre médical, et rassurez-vous j'ai bientôt terminé, j'adresserai en premier un remerciement spécial à notre Barman **RENAULT** pour son accueil au Gainzbar puis à la Grappe d'Or tout au long de mes 12 années d'études (et même avant cela…), je lui devais ces quelques lignes.

Les suivants et peut être mes plus chaleureux remerciements, à ma deuxième famille, à ceux qui ont été et seront toujours là, à ce groupe que nous nous sommes forgé et qui me remplit de fierté à chaque fois que je l'évoque ; merci à mes amis qui m'ont soutenu tout au long de ce parcours du combattant qu'ont été pour moi les études médicales. En citant à nouveau **LACOUR Emma** et **DENIS Axel**, mes compagnons de route, mais également **FOURNIER Antoine** et **LAVIGNE Quentin** pour nos soirées à la grappe, dans les bois ou que sais-je, souvent bien trop arrosées mais toujours incroyables ! A **CHARRIER Nathan** et **Morgane** pour leurs accueils constants lorsque l'envie de m'évader devenait trop forte, pour toutes ces soirées à parler d'autre chose que de médecine, merci. A **LAVIGNE Quentin**, **TUSSEAU Robin**, **DAMIEN Colas**, **Convul** et **Tropis** pour les tranches de rire incroyable sur le vocal. A **DAMIEN Colas**, **LEMERDY Claire**, **LAVIGNE Quentin** ou **DROUIN Jules**, ainsi qu'à **JH**, **HOTTO**, **Carole** et **Nico** pour les voyages au confins de l'imagination en GN qui m'ont permis de tenir jusqu'ici ! Et a **TUSSEAU Robin**, **DROUIN Jules**, **JH**, **LAVIGNE Quentin**, **CHARRIER Nathan** et tous les autres joueurs qui ont partagé avec moi une partie de JDR tout au long de ces années, merci !

REMERCIEMENTS

Thomas DAVID :

Enfin, je terminerais par remercier ma famille, soutenante à chaque instant, même lors de mes redoublements successifs et mes interminables rattrapages qui m'ont fait douter plus d'une fois. Merci à mes grands-parents **HUBERT Juliette** et **Jean-Pierre** et à **Tatie**, qui m'ont poussé et encouragé tôt vers les études. Merci à mes grands-parents **DAVID Marcel** et **Marie-Josephe** pour m'avoir fait connaitre le monde agricole et la vie à la campagne. Merci à mes oncles **HUBERT Pascal** et **Stéphane** ainsi que **DAVID Philippe** et également ma marraine **HUBERT Fabienne** pour leurs conseils et encouragements constants. Merci à mon premier professeur, mon père, **DAVID Dominique** qui m'a soutenu et me soutien encore ce jour, qui a su m'encourager à continuer mes études et qui a toujours répondu présent lors des galères de la vie, merci. Et un grand et énorme merci à mes frangins et frangines. Merci à **DAVID Pierre** et sa famille, pour la joie de vous retrouver, les vacances de déconnexion ensembles et mes deux merveilleuses nièces. Merci au presque docteur également **DAVID Etienne** pour nos soirées à déconner sur Lol, pour les conseils stratégiques et statistiques lors de la rédaction de ce papier, pour ton moral inébranlable merci. Merci à ma grande **DAVID Nadia**, pour tes choix avisés pour la planète et surtout l'espérance d'aller boire un verre ensemble sur Angers un soir, merci. Merci enfin au Big Boss **DAVID Kévin**, mon dernier frère et filleul, pour ton entièreté, ta force physique et morale, ta droiture et ta jovialité, merci !

Je terminerai par les deux seules personnes que je n'ai toujours pas citées.

A **HOUEIX Marie**, ma Caussenarde et mon amour, mais aussi ma béquille et bien souvent mon déambulateur ; pour notre rencontre magique dans les Causses il y a maintenant 8 ans ; pour ton appui sans faille et tes encouragements lors de ces longues années d'études et de la rédaction de ce papier ; pour nos week-end d'évasion et notre vie à deux me permettant de respirer à plein poumons ; pour ta capacité à simplifier tous mes problèmes et pour toutes les autres choses qui mériteraient bien plus que quelques lignes couchées sur du papier pour en faire ne serait-ce qu'une préface ; du fond du cœur, merci.

Et enfin à ma mère, **DAVID Béatrice**, qui m'aura connu médecin mais jamais connu docteur. Pour avoir été la maman la plus merveilleuse du monde pendant les 27 premières années de ma vie ; pour m'avoir fait gouter, par tes anecdotes d'infortune, la vie hospitalière et le monde des soignants ; pour avoir cultivé en moi le gout de la lecture et du travail bien fait ; pour m'avoir emmené en voiture chaque matin à la faculté lorsque les aller-et-retour quotidiens à vélo des 4 premiers mois de ma première année m'avait fait perdre 8 kilos, et cela même si tu t'étais pris une amende qui t'avait conduit au tribunal ; pour tes appels à la fin de chacune de mes journées d'examens ou rattrapage pour me demander comment ça s'était passé ; pour ton assistance sans faille lorsque mon moral était au plus bas à l'issu du concourt de l'internat ; et enfin pour ton combat acharné contre le cancer et ta lutte contre la souffrance même si elle a fini par l'emporter, et pour le cadeau que tu nous a fait d'avoir pu nous dire au revoir à chacun avant que l'on ne t'endorme en attendant la fin ; cette thèse t'es dédiée Maman parce sans toi je n'aurais probablement jamais pu aller aussi loin ; Merci infiniment.

Liste des abréviations

BEP	Brevet d'études professionnelles
BEPC	Brevet d'Études du Premier Cycle
BTS	Brevet de Technicien Supérieur
CAP	Certificat d'Aptitude Professionelle
DUT	Diplôme Universitaire de Technologie
IDEST	Infirmier Diplômé d'Etat
ABI	Ankle Brachial Index
MDA	Maison Départementale de l'Autonomie
PXE	PseudoXanthome Elastique
RQTH	Reconnaissance en Qualité de Travailleur Handicapé

Plan

Employment of Patients with Pseudoxanthoma Elasticum (PXE). A survey in the French reference center for PXE patients

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CRL and AP did the literature searches and created the questionnaire. CRL and TD made the CPP and ClinicalTrials reports, sent out the newsletters, made the patient calls, entered the data into Epidata and wrote the article. CR analysed the data. AP and LM supervised, corrected and validated all steps of the study.

RESUME

Background: *Pseudoxanthoma elasticum (PXE) is a rare, autosomal recessive disease characterized by progressive calcification and fragmentation of elastic fibers in connective tissue. PXE primarily affects the skin, retina and arterial walls. Given the age of onset and progression of the disease, the consequences of PXE affect a large number of working patients and are therefore likely to have an impact on their occupational career or job retention.*

Results: *99 patients with an average age of 43.8 years (sex ratio of 1 male to 2 females) were included in the study. Their complications were mainly visual and vascular. The study showed that the proportion of patients in our population whose employment was stopped or modified because of PXE was 8.1%. Dermatological complications did not appear to have had an impact on job retention. The analysis of our data shows that age, visual impairment and psychological impact of disease manifestations are major determinants of patients' job retention.*

Conclusions: *In the light of these data, prevention in occupational medicine and individual follow-up of these workers must be centered on visual problems and limb claudication. These data encouraged us to create a new consultation dedicated to the problem of maintaining employment for patients with PXE within the department of occupational medicine of Angers in close collaboration with the reference center.*

INTRODUCTION

Pseudoxanthoma elasticum (PXE) is a rare, autosomal recessive, inherited disease characterized by progressive calcification and fragmentation of elastic fibers in connective tissue. The manifestations are skin changes before the age of 20, claudication of the lower and upper limbs around the age of 40, increased risk of stroke, and finally a scotoma (central blindness) in the late 50s. PXE causes handicaps, sometimes leading to a loss of autonomy. Given the age of onset and progression of the disease, the consequences of PXE affect a large number of patients of working age and are therefore likely to have an impact on their occupational career and job retention. However, no study to date has evaluated the impact on the career path of patients with PXE.

The main objective of this study was therefore to evaluate the impact of PXE on the career pathway and job retention of patients suffering from this disease, as well as the medical reasons linked to job retention. The primary outcome was the percentage of job loss due to PXE. The secondary objectives were to evaluate the work experience of the consequences of PXE and the tools and actors involved in job retention as well as the results obtained, if any, as well as percentage of job losses due to the vascular impact, visual impact or skin impact. Finely, the study aimed to explore tools and stakeholders who are involved in job retention management.

This exploratory study will make it possible to verify the need for more in-depth studies and for personalized job retention support for patients.

MÉTHODES

1. Figure 1 : Flow diagram

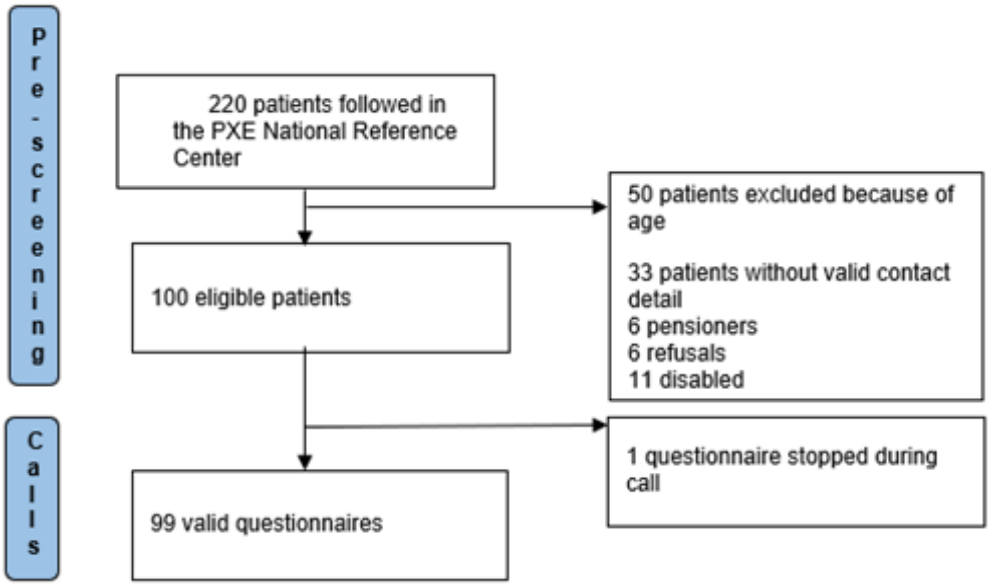

Figure 1 : Flow diagram.

The study was an epidemiological, descriptive and analytical, cross-sectional, monocentric study, carried out by a phone questionnaire in the French cohort of the PXE Reference Center in the Dermatology Department of the Angers University Hospital consisting of 220 patients. The inclusion criteria were to be diagnosed for a PXE and to be in to at working age (18 to 64 years old) or unemployed / retired for less than 2 years. The exclusion criteria were the presence of one or more physical or psychological comorbidity likely to have an impact on the career path and job retention; the poor understanding of the French language; the objection to participate in research. The main evaluation criterion was the employment status of the subjects as well as the medical reasons related to the loss of a job or a change of position or career path.

Eligible subjects were pre-selected by the study investigators on the basis of the medical file. A letter of information about the study was sent to all the pre-selected eligible patients, with the exception of patients who had expressed an objection to the processing of their data for research purposes.

The investigators contacted eligible patients by phone approximately 2 weeks after the mailing of the information letter. During this phone call, the investigator answered any questions the patient may have about the study and collected the patient's non-objection to participate in the study and their responses to the questionnaire.

This questionnaire was tested in order to verify its feasibility and clarity on 2 successive randomly selected patients. Medical data (age at diagnosis, age at referral to the Reference Center, organ damage and clinical manifestations present at the time of the survey, treatments, etc.) were collected from the medical file and the answers to the questionnaire were collected by phone on paper. Data were collected in a coded manner. A correspondence list has been kept, under the responsibility of the principal investigator.

The data collected by the questionnaire were entered by the investigators into an EPIDATA database implemented by the Clinical Research and Innovation Department of the CHU of Angers. Statistical analyses (Fisher’s exact test, Wilcoxon rank sum test and Pearson's Chi-squared test) were then carried out by a statistician from the Epidemiology in Occupational Health and Ergonomics laboratory in Angers.

RÉSULTATS

1. Results

The full study results are available on the attached tables in the appendix.

1.1. Population Characteristics and employment status (Tables A and B)

The study population consisted of 29 males (29%) to 70 females (71%). The mean age was 43.8 ± 11.9 years ([18 - 64 years], Table I). The full study results are available online as supplemental material.

1.2. Work and aesthetic damage (Table C)

Figure 2 : Pseudoxanthoma elasticum of the posterior lateral neck.

Concerning skin damage: 91 patients had cervical damage and 70 patients had antecubital damage.

Of the 98 patients, no patient had to change employment position because of aesthetic damage.

1.3. Work and Visual Acuity Decrease (Table D)

Of the 99 responders, 87 patients reported visual impairment. Forty nine patients reported a decrease in visual acuity related to PXE. In this population, near a quarter stated to be impacted by decreased visual acuity (change of employment position, change of company, exclusion of work or other).

1.4. Work and limb claudication (Table E)

Forty two patients reported having upper and/or lower limb claudication related to PXE; in 3 cases in the lower limbs, in 23 cases in the upper limbs and in 15 cases in both the lower and upper limbs. In this population, 5 people stated to be impacted by the claudication (change of workstation, change of company or exclusion from work).

1.5. Work and psychological impact (Table F)

Forty seven patients did not feel that the consequences of PXE were a source of stress in their work; 26 patients had some of this impression; 11 patients had this impression somewhat and 15 patients felt enormously this impression.

1.6. Job retention tools (Table G)

The different tools of job retention evaluated were the recognition as disabled workers (in French: "reconnaissance de la qualité de travailleur handicapé" or RQTH); disability pension; the adaptation of the workstation and the different actors of job retention mobilized and the results obtained.

Tableau I : Population characteristics.

			Number
Sex			
Male			29
Female			70
Not in employment			10
In employment			89
Dermatological involvement			
	No		1
	Yes		98
		Cervical	91
		Antecubital	70
Decreased Visual Acuity due to PXE			
	No		50
	Yes		49
PXE-related Claudication			
	No		57
	Yes		42
Consequences of PXE as a source of stress at work			
	Not		47
	A little		26
	Enough		11
	Enormously		15

2. Description of results

Statistical analyses revealed that unemployed patients were significantly older than employed ones (51.3 *versus* 43 years, p=0.022). Unemployed patients tended to be more likely to have decreased visual acuity (80% versus 20%, p=0.051) and to think that consequences of PXE are a source of stress at work (p=0.058) (Table II).

Tableau II : Comparison of patient's characteristics between unemployed and employed groups.

Characteristics	unemployed patients (N = 10) n (%); Mean (SD)	employed patients (N = 89) n (%); Mean (SD)	p-value[2]
Sex			0.152
Female	5 (50.0%)	65 (73.0%)	
Male	5 (50.0%)	24 (27.0%)	
Age (years)	51.3 (15.5)	43.0 (11.2)	**0.022**
Cervical dermatological involvement			>0.999
No	0 (0.0%)	8 (9.0%)	
Yes	10 (100.0%)	81 (91.0%)	
Antecubital dermatological involvement			>0.999
No	3 (30.0%)	26 (29.2%)	
Yes	7 (70.0%)	63 (70.8%)	
Visual acuity decrease due to PXE			**0.051**
No	2 (20.0%)	48 (53.9%)	
Yes	8 (80.0%)	41 (46.1%)	
PXE-related claudication			0.739
No	5 (50.0%)	52 (58.4%)	
Yes	5 (50.0%)	37 (41.6%)	
PXE causing stress at work			**0.058**
Not at all	3 (30.0%)	44 (49.4%)	
A little	1 (10.0%)	25 (28.1%)	
Enough	2 (20.0%)	9 (10.1%)	
Enormously	4 (40.0%)	11 (12.4%)	

[2]Fisher's exact test; Wilcoxon rank sum test

2.1. Tools for job retention

Tables I, J, K and L present the analyses concerning the various job retention tools.

2.1.1. RQTH

Among the patients who obtained the RQTH status (N=23), the proportion of people with a decrease in visual acuity related to PXE was very significantly higher compared to those who did not (91.3% *versus* 8.7%, p<0.001). Moreover, patients who obtained the RQTH status were significantly more often in employment was compared with those who did not (73.9% *versus* 26.1%, p=0.01).

2.1.2. Disability

Among the patients who perceived a disability pension (N=13), the proportion of people with visual impairment related to PXE was significantly higher than those without (84.6% *versus* 15.4%, p=0.008). Moreover, patients who perceived disability pension were very significantly more often in employment compared with those who did not (53.8% *versus* 46.2%, p< 0.001).

2.1.3. Support in maintaining employment

The patients who benefited from assistance in maintaining employment (N=25), were significantly more likely to have decreased visual acuity (80% *versus* 24% who did not, p<0.001). Moreover, they were significantly more often in employment compared with those who did not (76% versus 24%, p=0.015).

2.1.4. Adjustment of workstation

The patients who had received a workstation adjustment, either considered or undertaken (N=27), were significantly more likely to have decreased visual acuity (70.4% versus 29.6% who did not, p=0.020). The employment rate in this population tended to be higher but not significantly (81.5%).

These analyses showed the preponderant importance of the PXE visual consequences about maintaining employment in these patients. Furthermore, job retention tools such as RQTH,

disability pension, assistance in maintaining employment and workstation adjustment appeared to have a positive impact on job retention.

2.2. Additional analyses

Additional analyses can be found in Table H in the Supplemental online.

2.2.1. Visual impact

Regarding the visual impact of PXE, the complementary analyses indicated a statistical link between employment and the assessed impact of decreased visual acuity ($p=0.018$), to feel of taking more time than before to perform the same work tasks ($p<0.001$), mood disorders related to decreased visual acuity ($p=0.004$), to feel guilty or to have lowered professional esteem related to decreased visual acuity ($p=0.009$), to be able to drive ($p<0.001$); a decrease in central vision ($p=0.006$); to visit the Occupational Physician ($p<0.001$), and to be placed in work restrictions related to decreased visual acuity ($p=0.050$).

2.2.2. Psychological impact

Regarding PXE psychological impact, the complementary analyses indicated a statistical link between employment and having discussions with the employer about the PXE consequences ($p=0.012$).

DISCUSSION

Our study shows that employment stop or adjustment of work related to PXE complications concern 8.1% of the cohort. The complications were mainly related to visual impairment (6%) and vascular troubles (2%) while dermatological disorders did not lead to a change or a stop of work. Analyses showed that visual impact of PXE appeared to be the most important determinant of patient job retention.

It is important to specify that the sex ratio of 1 man to 2 women corresponds to the usual sex distribution in PXE cohorts. The proportion of people at work in our study seems to be higher than the national average (about 67.3% for people aged 15 to 64)[12], but these two values are hardly comparable in the face of deferential selection criteria (our population was selected from 18 to 64 years old and we excluded patients on disability). The distribution of the different sectors of activity (industrial, construction, commercial and services) seems roughly comparable to that of the general French population [12.] These elements make us think that our population, although small in size, is representative of the general population of PXE patients.

The slow and progressive evolution remains one of the main characteristics of PXE. This means that the most affected patients are globally the oldest. It is therefore not surprising to find that age is one of the job retention determinants in this population. But this long-time interval can also be seen as an opportunity. By anticipating and informing the different work actors (patient, Occupational Physician or General Practitioner) about the inherent risks related to this disease and its evolution, and especially about the functional impact on work, could have an impact on job retention and working life.

For example, on the occasion of their follow-up in the referral center, we decided to give patients an information letter about PXE for their Occupational Physician. This letter is expected to allow regular early screening for visual and vascular disorders to be carried out from the beginning of the career. That would allow improving dialogue between the patient and its Occupational Physician to consider job adjustments from the symptom’s onset. That could also minimize the impact on patients' stress at work.

As an extension of our study, we set up a specific consultation in the Department of Occupational Medicine in order to make a global review of the different tolls and stakeholders dedicated to job retention during visits to the PXE reference center. It would also make it possible to detect situations at risk of job retention problems when the link with the Occupational Physician has not yet been made. A prospective study based on the same questionnaire after the consultation has been set up and will be carried out to see the consultation relevance.

The lack of literature on PXE and job retention calls for a more powerful study on this topic and raises questions about a possible lack of support for patients in maintaining employment. It could be interesting to imagine that, at the national level, all rare diseases reference centers could produce this kind of standard letter for the Occupational Physician through the patient. This could allow occupational Physician to better assess the potential impact of rare diseases at work.

One of the limitations of this study was related to its retrospective cross-sectional study design. The exploratory study design implies that no definite conclusions can be made. Another limitation was the exclusion of patients who had not worked for more than two years. Our main idea was that we needed a working population to assess the impact of PXE on employment. However, we excluded from our research a number of patients who were not considered "active" in the legal sense of the term, but who nevertheless had problems in maintaining employment because of PXE. At least ten of the patients that we excluded from the study perceived a disability pension related to PXE. It would be wise to include them in future studies to better understand the complications that led to their disability and the follow-up they received.

CONCLUSION

This work illustrates the opportunity for Occupational Physicians to be involved in the care management of rare diseases patients.

In the light of these original data, it is certain that prevention in occupational medicine and in the individual follow-up of these workers must be centered on their disease experience in their job, and more especially on visual and claudication disorders, which seem to emerge as the two determining factors of job retention in these patients.

These results must be put into perspective because of the selection of our working population and that must be taken into account for future studies.

In any case, these results encourage the development of studies related to the job retention issue in patients with PXE including creation of consultations dedicated to job retention for PXE patients but also for others rare diseases.

BIBLIOGRAPHIE

[1] Chassaing N, Martin L, Calvas P, Le Bert M, Hovnanian A, Pseudoxanthoma elasticum: a clinical, pathophysiological and geneticupdate including 11 novel ABCC6 mutations. J Med Genet. 2005 Dec;42(12):881-92 2005 May 13

[2] Questionnaire pluridiscuplinaire Lombaction du Centre de consultation de Pathologie Professionnelle et Santé au travail (CHU d'Angers)

[3] Gupta MA, et al Age and gender differences in the impact of psoriasis on quality of life. Int J Dermatol. 1995. PMID: 8537157

[4] Little AC, Roberts CS Evolution, appearance, and occupational success. Evol Psychol. 2012;10(5):782-801

[5] The Impact of Vision Loss In : Making Eye Health a Population Health Imperative: Vision for Tomorrow. 2016 Sept 15

[6] Sarah E Blaylock and Al. Understanding the occupational performance experiences of individuals with low vision. British Journal Of Occupationnal Therapy Volume: 78 issue: 7, page(s): 412-421 May 29, 2015.

[7] Josée Duquette And Al ; Occupational performance in the basic and instrumental daily activities of persons with low vision who received rehabilitation services. British Journal Of Occupationnal Therapy Volume: 82 issue: 8, page(s): 457-465 November 14 2018

[8] Chua B, Mitchell P. Consequences of amblyopia on education, occupation, and long term visionloss. Br J Ophthalmol. 2004 Sep;88(9):1119-21.

[9] Anne-Elisabeth Chaumet-Riffaud and Al ; Handicap visuel et maintien en emploi : étude dans la rétinite pigmentaire, Revue Francophone d'Orthoptie Volume 10, Issue 1, January–March 2017, Page 24

[10] Huo Yung Kai S, Ferrières J, Carles C, Turpin M, Lapébie FX, Dutheil F, Bura-Rivière A, Esquirol Y Lower limb venous and arterial peripheral diseases and work conditions: systematic review. Occup Environ Med. 2021 Jan;78(1):4-14.

[11] P Lacroix 1, V Aboyans, C Boissier, L Bressollette, P Léger Validation of a French translation of the Edinburgh claudication questionnaire among general practitioners' patients. Arch Mal Coeur Vaiss. 2002 Jun;95(6):596-600.

[12] Emploi, chômage,revenus du travail. Insee Références, édition 2022

DEFINITIONS

Visual Acuity: Quantity measuring the capacity of the eye to discriminate two distinct points, according to the distance separating them and the distance of the observer. Expressed in tenths.

Health Insurance: Health Insurance is a branch of the French Social Security system that guarantees health protection to all insured persons. In order to ensure its proper functioning, each insured person contributes and in return benefits from coverage according to a defined scale. The scope of action of the Health Insurance includes:

Sickness, maternity and disability: it covers all or part of your health expenses, pays daily allowances in case of work stoppage and a pension in case of disability.

Occupational illnesses, work accidents and death: it pays for treatment, provides daily allowances in the event of work stoppage and an annuity to the beneficiaries in the event of death.

BEP: Abbreviation for Brevet d'études professionnelles (meaning vocational studies certificate), a French diploma.

BEPC: Abbreviation for Brevet d'Études du Premier Cycle (meaning first cycle certificate of education), a diploma obtained in France until 1977 now known as the diplôme national du brevet

BTS: Abbreviation for Brevet de Technicien Supérieur (meaning superior technician certificate), a french diploma.

CAP: Abbreviation for Certificat d'Aptitude Professionelle (meaning certificate of professional aptitude), a French diploma.

Cap Emploi: financial companies carrying out a public service mission. They are in charge of preparing, accompanying, monitoring and maintaining the employment of people with

disabilities. They welcome and support more than 220,000 people with disabilities and more than 150,000 employers each year.

Low Vision Center: regional follow-up care and visual and auditory rehabilitation service.

DUT: Abbreviation for Diplome Universitaire de Technologie (meaning University Diploma of Technology), a French diploma.

Idest: Abbreviation for Infirmier Diplômé d'Etat de Santé au Travail (meaning State Registered Occupational Health Nurse). This nurse has undergone specific training in occupational health. He is bound by medical secrecy. He works in cooperation with the occupational physician and under his authority and participates in the individual health monitoring of employees

Disability: Disability in the sense of the Health Insurance is a recognition of the reduction of an individual's work capacity by the French state due to his or her health condition. This reduction in work capacity is compensated by the payment of specific state allowances to supplement the employee's reduced salary.

ABI: Ankle Brachial Index is the ratio of systolic blood pressure (SBP) measured at the ankle to brachial systolic blood pressure.

MDA : Abbreviation for Maison Départementale de l'Autonomie (meaning departmental house of autonomy), a departmental public structure whose mission is to welcome, inform, support and advise disabled people and their families, and to raise awareness of disabilities among all citizens.

Occupational physician: Occupational medicine is a clinical specialty, and is the branch of medicine that is focused on keeping individuals well at work, both mentally and physically.

Reclassification, internal or external: action of the employer aiming at transferring an employee to a job different from the one he/she occupies. Either to another different position within the company (within the national territory), in which case we speak of internal reclassification. Or

to a different position within another company or on the job market, this is called external redeployment.

RQTH: Abbreviation for Reconnaissance en Qualité de Travailleur Handicapé (meaning Recognition as a disabled worker), French administrative decision that grants disabled persons a status that allows them to benefit from specific assistance, particularly in terms of employment and/or job accommodation. It is issued by the Maison Départementale de l'Autonomie.

Skill assessment: analysis of the professional and personal skills, aptitudes and motivations of an employee, carried out in support of a professional development and/or training project.

Therapeutic part-time work: A special form of return to work after an absence due to illness or accident, whether occupational or non-occupational, designed to improve the employee's state of health or when the employee is undergoing re-education or vocational rehabilitation in order to find a job compatible with his or her state of health. It can also be implemented after a return to work and be requested by the employee. It is prescribed by the attending physician and organized jointly with the employer and the occupational physician.

LISTE DES FIGURES

LISTE DES TABLEAUX

ANNEXES

TABLE DES MATIERES

ANNEXES

Table A : Population Characteristic.

			Number	Average age	Average age at diagnosis
Male			29	45,4	28,9
	Marital status				
		Single	4		
		In a couple	8		
		Had contracted a civil union	5		
		Married	11		
		Divorced	1		
		Widowed	0		
	School level				
		Post-graduate degree, doctorate, graduate school, engineer	3		
		2nd cycle university diploma	5		
		1st cycle university diploma, BTS, DUT or equivalent, level BAC+2	8		
		General, technological or professional baccalaureate or equivalent	4		
		CAP, BEP or diploma of the same level	9		
		Brevet des collèges, BEPC	0		
		Primary school certificate, no diploma	0		
Female			70	43,1	23,1
	Statut matrimonial				
		Single	17		
		In couple	16		
		Had contracted a civil union	6		
		Married	26		
		Divorced	5		
		Widowed	0		
	Niveau scolaire				
		Post-graduate degree, doctorate, graduate school, engineer	4		
		2nd cycle university diploma	23		
		1st cycle university diploma, BTS, DUT or equivalent, level BAC+2	21		

Table B : Employment Status.

Not in employment	10					
	Status					
		Unemployed	3			
		Retired	4			
		Other	3			
In Employment	89					
	Sectors					
		Agricultural	3			
		Industry	14			
		Construction	8			
		Commerce	11			
		Services	53			
	Duration of employment					
		Less than 1 year	18			
		Between 1 and 5 years	13			
		Between 5 and 10 years	22			
		More than 10 years	36			
	Type of contract					
		Permanent contracts	50			
		Fixed-term contracts	7			
		Civil servants	22			
		Self-employed	8			
		Temporary contract	1			
		Other	1			

Table C : Aesthetic Damage.

Dermatological involvement						
	No	1				
	Yes	98				
		Cervical	91			
		Antecubital	70			
Contact with the public						
	No	27				
	Yes	71				
		Frequency				
		Daily	53			
		Multi-weekly	53			
		Weekly	3			
		Less frequent	5			
Aesthetic damage						
	No	48				
	Yes	50				
		Judged as:				
			Very important	6		

			Somewhat important	21		
			Not very important	17		
			Not at all important	6		
				Standard deviation	Min	Max
		Since (average)	20,3	11,6	4	50
Cosmetic surgery						
No	66					
Yes	32					
	Considered	23				
	Carried out	9				
		Since				
			Less than 6 months	2		
			Less than 1 year	7		
			Less than 5 years	10		
			Less than 10 years	5		
			More than 10 years	8		
Caused discomfort						
	Never	64				
	Rarely	13				
	Often	11				
	Always	10				
		To:				
			Colleagues	22		
			Hierarchy	12		
			Public	22		
			Other	2		
Remarks						
	Never	76				
	Rarely	18				
	Often	4				
	Always	0				
		From :				
			Colleagues	17		
			Hierarchy	5		
			Public	9		
			Other	1		
Wearing clothing						
	Never	73				
	Rarely	3				
	Often	4				
	Always	18				
Renunciation						
	No	97				
	Yes	1				

Change of position						
	No	98				
	Yes	0				
Felt						
	Stigmatization	6				
	Unable to go unnoticed	8				
	Embarrassment	22				
	Malaise	7				
	Looks of curiosity	40				
	Devisagement	20				
	Need isolation	5				
	Other	0				

Table D : Visual Acuity Decrease.

Decreased Visual Acuity due to PXE							
	No	50					
	Yes	49					
		Since:					
			Average	Standard deviation	Min	Max	
			11,7	11,9	1	50	
		Degrees of severity					
			Judged as:				
				Very important	21		
				Somewhat important	12		
				Not very important	15		
				Not at all important	1		
		Device					
		No	7				
		Yes	42				
			Type :				
				Tablet	9		
				Magnifying glass	11		
				Glasses	41		
				Other	5		
		Impact of decreased visual acuity on work					
			Not	8			
			Yes	41			
				Judged as			
					Very important	16	

					Somewhat important	13	
					Not very important	12	
					Not at all important	0	
		Visual fatigue at work					
			Not	12			
			Yes	37			
				Yes physical	32		
				Yes intellectual	28		
					For:		
						Work on screens	30
						Reading documents	30
						Careful task completion	23
						Lack of lighting	30
						Other	3
					At the end of:		
						Average (min)	
					139,9	132,7	
		Impression of taking longer					
			Not at all	17			
			A little	12			
			Enough	4			
			Enormously	16			
		Signs associated with decreased visual acuity					
			Attention deficit/concentration disorder	23			
			Mood disorder	26			
			Loss of quality of work	22			
			Increased risks at work	16			
			Other	2			
		Guilt, degradation of professional esteem related the decrease in visual acuity					
			Not	34			
			Yes	15			
		Consultation in a Low Vision Center					
			Not	40			
			Yes	9			
				Proposed solutions			
					Equipment	5	

					Workstation adjustment	3	
					Rehabilitation	4	
					Other	0	
		Special situations					
			Not	37			
			Yes	12			
				Change of position	7		
				Change of company	1		
				Exclusion from the world of work	5		
				Other	1		
		Consultation with an occupational physician					
			Not	35			
			Yes	14			
				Proposed solutions:			
					Equipment	6	
					Workstation adjustment	8	
					Internal reclassification	2	
					External reclassification	0	
					Restrictions	4	
					Dismissal for incapacity	5	
					Other	1	
		Restrictions on the workstation					
			Not	45			
			Yes	4			
		Workstation adjustment in connection with the decreased visual acuity					
			Not	32			
			Yes	17			

Table E : PXE-related Claudication.

PXE-related Claudication					
Not	57				
Yes	42				
	Members				
		Superior	3		
		Lower	23		
		Superior and lower	15		
	For what type of activity				
		Prolonged standing	22		
		Prolonged walking	29		

		Work with the arms above the plane of the shoulders	18		
		Repetitive gestures	25		
		Hard work	21		
	Impact on work deemed as:				
		Very important	8		
		Somewhat important	8		
		Not very important	13		
		Not at all important	13		
	Mutual aid from colleagues				
		Not	31		
		Yes	11		
	Workstation adjustment in connection with lameness				
		Not	24		
		Yes	18		
			Directed by:		
				Ergonomist	0
				Occupational Physician	4
				Cap Emploi	0
				Hierarchy	0
				Idest	0
				Other	15
	Workstation adjustment due to claudication				
		Not	37		
		Yes	5		
			Become:		
				Change of workstation	3
				Change of company	1
				Exclusion from work	1
				Other	0

Table F : Psychological impact.

Consequences of PXE as a source of stress at work		
	Not	47
	A little	26
	Enough	11
	Enormously	15
Consequences of PXE increasing stress already present		
	Yes	50
	Not	49
Criticism at work in relation to one of the consequences of PXE		
	Never	95
	Rarely	1
	Often	1
	Always	2

Conflicts with colleagues related to one of the consequences of PXE		
	Never	93
	Rarely	4
	Often	1
	Always	1
Discussions with the employer regarding one of the consequences of the PXE		
	Never	66
	Rarely	27
	Often	3
	Always	3
Refusal of employment		
	Not	92
	Yes	7
Fear of lost wages		
	Never	62
	Rarely	7
	Often	8
	Always	22
Fear of job loss		
	Never	55
	Rarely	11
	Often	9
	Always	24
Dismissal due to PXE		
	Not	91
	Yes	8
Stopping studies		
	Not	96
	Yes	3

Table G : Job retention tools.

RQTH					
	Not	76			
		Reasons			
			Misunderstanding	33	
			Uselessness	53	
			Patient refusal	1	
			MDA Refusal	0	
			Other	1	
	Yes	23			
		Average recognition time	7		
Disability					
	Not	86			
	Yes	13			
		Category 1	8		
		Category 3	4		

		Category 3	1		
		Average duration of disability	6,4		
Station layout					
	Not	72			
		Reasons			
			Uselessness	55	
			Employer refusal	0	
			Impossible	10	
			Other	0	
	Yes	27			
		Considered	5		
			Since		
				Less than 6 me	3
				More than 6 months	2
		Undertaken	22		
			Since		
				Less than a year	8
				Less than 5 years	9
				Less than 10 years	1
				More than 10 years	4
			Type of development		
				Ergonomic	17
				Organizational	17
				Other	7
		Link to PXE			
		No	10		
		Yes	17		
			Problematic		
				Aesthetic	0
				Visual	15
				Claudication	5
				Psychic	0
				Other	1
Support for job retention					
	No	74			
	Yes	25			
		Since			
			Less than 6 months	6	
			Less than a year	6	
			Less than 5 years	9	
			Less than 10 years	3	
			More than 10 years	1	
		Actors mobilized			
			LabourDoctor	21	
			Médecin de l'assurance maladie	9	
			DRH	9	

			MDA	8	
			Ergonomist	6	
			Other	5	
		Results			
			Station layout	1	
			Internal redeployment	2	
			Outplacement	0	

Table H : Further Analysis.

Characteristic	No, N = 10[1]	Yes, N = 89[1]	p-value[2]
Sex			0.152
Female	5 (50.0%)	65 (73.0%)	
Male	5 (50.0%)	24 (27.0%)	
Age (years)	51.3 (15.5)	43.0 (11.2)	**0.022**
Cervical dermatological involvement			>0.999
No	0 (0.0%)	8 (9.0%)	
Yes	10 (100.0%)	81 (91.0%)	
Antecubital dermatological involvement			>0.999
No	3 (30.0%)	26 (29.2%)	
Yes	7 (70.0%)	63 (70.8%)	
Visual impairment			0.605
No	0 (0.0%)	12 (13.5%)	
Yes	10 (100.0%)	77 (86.5%)	
Visual acuity decrease due to PXE			**0.051**
No	2 (20.0%)	48 (53.9%)	
Yes	8 (80.0%)	41 (46.1%)	
Perceived degree of severity of VAD			0.157
Very important	5 (50.0%)	20 (22.5%)	
Somewhat important	2 (20.0%)	11 (12.4%)	
Not very important	1 (10.0%)	17 (19.1%)	
Not at all important	2 (20.0%)	41 (46.1%)	
Impact of decreased visual acuity on work			0.097
No	3 (30.0%)	53 (59.6%)	
Yes	7 (70.0%)	36 (40.4%)	

Characteristic	No, N = 10[1]	Yes, N = 89[1]	p-value[2]
Impact: significance			**0.018**
Very important	6 (85.7%)	10 (27.8%)	
Somewhat important	1 (14.3%)	14 (38.9%)	
Not very important	0 (0.0%)	12 (33.3%)	
Missing data	3	53	
Perceived fatigue related to visual efforts at work			0.318
No	3 (30.0%)	46 (51.7%)	
Yes	7 (70.0%)	43 (48.3%)	
Physical fatigue related to visual efforts at work			0.324
No	0 (0.0%)	11 (25.6%)	
Yes	7 (100.0%)	32 (74.4%)	
Missing data	3	46	
Intellectual fatigue related to visual efforts at work			>0.999
No	2 (28.6%)	14 (32.6%)	
Yes	5 (71.4%)	29 (67.4%)	
Missing data	3	46	
Feeling like it's taking longer to do work tasks than before because of BAV			**<0.001**
Not at all	3 (30.0%)	63 (70.8%)	
A little	0 (0.0%)	13 (14.6%)	
Enough	0 (0.0%)	4 (4.5%)	
Enormously	7 (70.0%)	9 (10.1%)	
Attention deficit/concentration disorder			0.695
No	7 (70.0%)	68 (77.3%)	
Yes	3 (30.0%)	20 (22.7%)	
Missing data	0	1	
Mood disorder			**0.004**
No	3 (30.0%)	68 (77.3%)	
Yes	7 (70.0%)	20 (22.7%)	
Missing data	0	1	
Loss of quality of work			0.225
No	6 (60.0%)	70 (79.5%)	
Yes	4 (40.0%)	18 (20.5%)	

Characteristic	No, N = 10[1]	Yes, N = 89[1]	p-value[2]
Missing data	0	1	
Increased risk at work			0.371
No	7 (70.0%)	74 (84.1%)	
Yes	3 (30.0%)	14 (15.9%)	
Missing data	0	1	
Guilt, degradation of professional esteem related the decrease in visual acuity			**0.009**
No	5 (50.0%)	78 (87.6%)	
Yes	5 (50.0%)	11 (12.4%)	
Driving possible ?			**<0.001**
No	7 (70.0%)	7 (8.0%)	
Yes	3 (30.0%)	77 (88.5%)	
Non responding	0 (0.0%)	3 (3.4%)	
Missing data	0	2	
Consultation in a Low Vision Center			**0.006**
No	6 (60.0%)	82 (94.3%)	
Yes	4 (40.0%)	5 (5.7%)	
Missing data	0	2	
Consultation with an occupational physician			**<0.001**
No	4 (40.0%)	80 (89.9%)	
Yes	6 (60.0%)	9 (10.1%)	
Restrictions on the workstation			**0.050**
No	8 (80.0%)	87 (97.8%)	
Yes	2 (20.0%)	2 (2.2%)	
Workstation adjustment in connection with the decreased visual acuity			0.399
No	7 (70.0%)	73 (82.0%)	
Yes	3 (30.0%)	16 (18.0%)	
Cardiovascular impairment			0.507
No	3 (30.0%)	40 (44.9%)	
Yes	7 (70.0%)	49 (55.1%)	
PXE-related claudication			0.739
No	5 (50.0%)	52 (58.4%)	
Yes	5 (50.0%)	37 (41.6%)	

Characteristic	No, N = 10[1]	Yes, N = 89[1]	p-value[2]
Do you feel that the consequences of PXE are causing stress at work?			**0.058**
Not at all	3 (30.0%)	44 (49.4%)	
A little	1 (10.0%)	25 (28.1%)	
Enough	2 (20.0%)	9 (10.1%)	
Enormously	4 (40.0%)	11 (12.4%)	
Have the consequences of PXE generated or increased your level of stress at work?			0.200
No	3 (30.0%)	47 (52.8%)	
Yes	7 (70.0%)	42 (47.2%)	
Have you been criticized for taking too much time out of your workday for medical appointments?			>0.999
Never	10 (100.0%)	85 (95.5%)	
Rarely	0 (0.0%)	1 (1.1%)	
Often	0 (0.0%)	1 (1.1%)	
Always	0 (0.0%)	2 (2.2%)	
Have you had conflicts with colleagues because of the aesthetic, visual or other consequences of PXE?			0.111
Never	8 (80.0%)	85 (95.5%)	
Rarely	2 (20.0%)	2 (2.2%)	
Often	0 (0.0%)	1 (1.1%)	
Always	0 (0.0%)	1 (1.1%)	
Have the consequences of the PXE resulted in discussions with your employer/supervisor?			**0.012**
Never	4 (40.0%)	62 (69.7%)	
Rarely	3 (30.0%)	24 (27.0%)	
Often	2 (20.0%)	1 (1.1%)	
Always	1 (10.0%)	2 (2.2%)	
Have you been denied a job/promotion due to the impact of PXE?			0.147
No	8 (80.0%)	84 (94.4%)	
Yes	2 (20.0%)	5 (5.6%)	
Are you/Where you concerned that the consequences of the PXE will result in lost wages?			0.197
Not at all	5 (50.0%)	57 (64.0%)	
A little	0 (0.0%)	7 (7.9%)	

Characteristic	No, N = 10[1]	Yes, N = 89[1]	p-value[2]
Enough	0 (0.0%)	8 (9.0%)	
Enormously	5 (50.0%)	17 (19.1%)	
Are you/Where you afraid of losing your job as a result of the PXE?			0.070
Not at all	4 (40.0%)	51 (57.3%)	
A little	0 (0.0%)	11 (12.4%)	
Enough	0 (0.0%)	9 (10.1%)	
Enormously	6 (60.0%)	18 (20.2%)	
Have you been terminated as a result of the PXE ?			**0.003**
No	6 (60.0%)	85 (95.5%)	
Yes	4 (40.0%)	4 (4.5%)	
Did you have to stop your studies/training because of the consequences of PXE?			0.276
No	9 (90.0%)	87 (97.8%)	
Yes	1 (10.0%)	2 (2.2%)	

[1]n (%); Mean (SD)

[2]Fisher's exact test; Wilcoxon rank sum test

Table I : RQTH and PXE.

Characteristic	No, N = 76[1]	Yes, N = 23[1]	p-value[2]
Cervical dermatological involvement			0.677
No	7 (9.2%)	1 (4.3%)	
Yes	69 (90.8%)	22 (95.7%)	
Antecubital dermatological involvement			0.901
No	23 (30.3%)	6 (26.1%)	
Yes	53 (69.7%)	17 (73.9%)	
Visual acuity decrease due to PXE			**<0.001**
No	48 (63.2%)	2 (8.7%)	
Yes	28 (36.8%)	21 (91.3%)	
PXE-related claudication			0.187
No	47 (61.8%)	10 (43.5%)	
Yes	29 (38.2%)	13 (56.5%)	
Currently employed ?			**0.010**

Characteristic	No, N = 76[1]	Yes, N = 23[1]	p-value[2]
No	4 (5.3%)	6 (26.1%)	
Yes	72 (94.7%)	17 (73.9%)	

[1]n (%)

[2]Fisher's exact test; Pearson's Chi-squared test

Table J : Legal Disability and PXE.

Characteristic	Disability, N = 13[1]	No disability, N = 86[1]	p-value[2]
Cervical dermatological involvement			0.068
No	3 (23.1%)	5 (5.8%)	
Yes	10 (76.9%)	81 (94.2%)	
Antecubital dermatological involvement			0.193
No	6 (46.2%)	23 (26.7%)	
Yes	7 (53.8%)	63 (73.3%)	
Visual acuity decrease due to PXE			**0.008**
No	2 (15.4%)	48 (55.8%)	
Yes	**11 (84.6%)**	38 (44.2%)	
PXE-related claudication			0.068
No	4 (30.8%)	53 (61.6%)	
Yes	9 (69.2%)	33 (38.4%)	
Currently employed ?			**<0.001**
No	6 (46.2%)	4 (4.7%)	
Yes	7 (53.8%)	82 (95.3%)	

[1]n (%)

[2]Fisher's exact test

Table K : Support in maintaining employment and PXE.

Characteristic	No, N = 74[1]	Yes, N = 25[1]	p-value[2]
Cervical dermatological involvement			**0.023**
No	3 (4.1%)	5 (20.0%)	
Yes	71 (95.9%)	20 (80.0%)	
Antecubital dermatological involvement			0.550
No	20 (27.0%)	9 (36.0%)	
Yes	54 (73.0%)	16 (64.0%)	

Characteristic	No, N = 74[1]	Yes, N = 25[1]	p-value[2]
Visual acuity decrease due to PXE			**<0.001**
No	45 (60.8%)	5 (20.0%)	
Yes	29 (39.2%)	20 (80.0%)	
PXE-related claudication			>0.999
No	43 (58.1%)	14 (56.0%)	
Yes	31 (41.9%)	11 (44.0%)	
Currently employed ?			**0.015**
No	4 (5.4%)	6 (24.0%)	
Yes	70 (94.6%)	19 (76.0%)	

[1]n (%)

[2]Fisher's exact test; Pearson's Chi-squared test

Table L : Workstation adjustment and PXE.

Characteristic	No, N = 72[1]	Considered or Undertaken N = 27[1]	p-value[2]
Cervical dermatological involvement			0.208
No	4 (5.6%)	4 (14.8%)	
Yes	68 (94.4%)	23 (85.2%)	
Antecubital dermatological involvement			0.430
No	19 (26.4%)	10 (37.0%)	
Yes	53 (73.6%)	17 (63.0%)	
Visual acuity decrease due to PXE			**0.020**
No	42 (58.3%)	8 (29.6%)	
Yes	30 (41.7%)	19 (70.4%)	
PXE-related claudication			0.633
No	43 (59.7%)	14 (51.9%)	
Yes	29 (40.3%)	13 (48.1%)	
Currently employed ?			0.130
No	5 (6.9%)	5 (18.5%)	
Yes	67 (93.1%)	22 (81.5%)	

[1]n (%)

[2]Fisher's exact test

REGNAULT-LEUGER Charlotte et DAVID Thomas

Problématiques de maintien en emploi chez les patients atteints de Pseudoxanthome Élastique

RÉSUMÉ

Contexte : Le pseudoxanthome élastique (PXE) est une maladie rare, autosomique récessive, caractérisée par une calcification et une fragmentation progressives des fibres élastiques du tissu conjonctif. Le PXE affecte principalement la peau, la rétine et les parois artérielles. Compte tenu de l'âge d'apparition et de la progression de la maladie, les conséquences du PXE touchent un grand nombre de patients actifs et sont donc susceptibles d'avoir un impact sur leur carrière professionnelle ou leur maintien dans l'emploi.

Résultats : 99 patients d'un âge moyen de 43,8 ans (sex ratio de 1 homme pour 2 femmes) ont été inclus dans l'étude. Leurs complications étaient principalement visuelles et vasculaires. L'étude a montré que la proportion de patients de notre population dont l'emploi a été arrêté ou modifié en raison du PXE était de 8,1%. Les complications dermatologiques ne semblent pas avoir eu d'impact sur le maintien dans l'emploi. L'analyse de nos données montre que l'âge, la déficience visuelle et l'impact psychologique des manifestations de la maladie sont des déterminants majeurs du maintien en emploi des patients.

Conclusions : A la lumière de ces données, la prévention en médecine du travail et le suivi individuel de ces travailleurs doivent être centrés autour des problèmes visuels et de la claudication des membres. Ces données nous ont incités à créer une nouvelle consultation dédiée à la problématique du maintien dans l'emploi des patients atteints de PXE au sein du service de pathologie professionnelle d'Angers en étroite collaboration avec le centre de référence.

Mots-clés : travail, emploi, maintien en emploi, impact de la maladie, maladie cardiovasculaire, maladie génétique, maladie des tissus conjonctifs, maladies dermatologiques

Employment of Patients with Pseudoxanthoma Elasticum (PXE). A survey in the French reference center for PXE patients

ABSTRACT

Background: Pseudoxanthoma elasticum (PXE) is a rare, autosomal recessive disease characterized by progressive calcification and fragmentation of elastic fibers in connective tissue. PXE primarily affects the skin, retina and arterial walls. Given the age of onset and progression of the disease, the consequences of PXE affect a large number of working patients and are therefore likely to have an impact on their occupational career or job retention.

Results: 99 patients with an average age of 43.8 years (sex ratio of 1 male to 2 females) were included in the study. Their complications were mainly visual and vascular. The study showed that the proportion of patients in our population whose employment was stopped or modified because of PXE was 8.1%. Dermatological complications did not appear to have had an impact on job retention. The analysis of our data shows that age, visual impairment and psychological impact of disease manifestations are major determinants of patients' job retention.

Conclusions: In the light of these data, prevention in occupational medicine and individual follow-up of these workers must be centered around visual problems and limb claudication. These data encouraged us to create a new consultation dedicated to the problem of maintaining employment for patients with PXE within the occupational pathology department of Angers in close collaboration with the reference center.

Keywords : work, job, employment retention, disease impact, cardiovascular diseases, genetic diseases, connective tissue diseases, skin diseases

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