Exploring Pain in Pachyonchia Congenita

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Introduction

Pachyonchia Congenita (PC) is a rare genetic skin disorder that causes thickening of the skin, nails, and mucous membranes. The pain associated with PC is often described as neuropathic, but the scale of this pain and its impact on quality of life (QoL) is not well understood. In this study, we aimed to explore the nature, severity, and impact of pain in a cohort of PC patients, and to measure the prevalence of neuropathic pain (NeP) within the four subtypes of PC.

Methods

1. A cross-sectional study of patients in the USA with full IRB ethics approval
2. 30 patients with a confirmed diagnosis of PC aged 18-80 included
3. Information was gathered using: 1. painDETECT™ questionnaire (pD): a validated tool for predicting neuropathic pain [3] 2. Clinical examination by an experienced clinician
4. EQ-5D: a standardised patient reported outcome measure of health status [4]
5. From EQ-SDs, a summary QoL score (EQ-5D index) was derived using US reference data [5]

Results

• The mean QoL-based health state observed has a severity that the average US citizen would forfeit one-third of remaining lifespan to avoid.

Discussion

• Pain is a primary symptom in PC and NeP is a prominent component of this pain
• K17 subtypes have the lowest EQ-5D score (therefore the highest burden of disease) and the highest NeP component.

Conclusion

It is hoped that this increased awareness of the burden of neuropathic pain in Pachyonchia Congenita will allow for more targeted pain therapies and improved quality of life.

References


Declaration

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