Pigmentosus xeroderma group D: A clinical and genetic study of 19 Japanese cases

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Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disorder of DNA repair in which the ability to repair damage caused by ultraviolet light with an incidence of 1: 22,000 in Japan. From the clinical symptoms, XP is classified into 3 types, such as XP cutaneous disease, XP neurological disease, and XP/Cockayne syndrome (CS) complex. Genetically, XP is classified into 22 subtypes. Although it is a very rare disease (VDRPA is the majority (55%) and XPV follows it (25%) in Japanese XP patients. However, there are many patients with XP cutaneous disease in Japan. In spite of the phenotypic and geographic differences, Japanese XP cases have not been adequately studied. So, we conducted the clinical, epidemiological, and genetic studies on 19 Japanese XP cases attended in our institution from 1998 to 2017 using medical and letters of laboratory data. The mean age of which a final diagnosis of XPD was 35.6 years old and all patients had a history of sunburn. In the clinical analysis, there were 15 cases of XPD cutaneous disease, 1 case of XP neurological disease, and 3 cases of XP/CS complex. Twelve of the 19 cases had complications of skin malignancies. The age at onset of skin cancer was 44.4 years for cutaneous disease, 12 years for neurological disease, and 2 years for XPCS complex. By the genetic analysis, we assumed that R601L was associated with cutaneous disease, G47R was associated with severe XP/CS complex, and R601G was associated with mild XP/CS complex. In conclusion, we have conducted the clinical, epidemiological, and genetic studies on 19 Japanese XP cases. The type of XPD cutaneous disease is dominant in Japanese XP cases. And genetic type (phenotype) of genotype association is implied in Japanese XPD patients from the confirmed gene mutation.

The impact of pemphigus on health-related quality of life: First results with the EQ-5D questionnaire

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The objective of our study was to evaluate health-related quality of life (HRQoL) in pemphigus patients using the EQ-5D and to analyse its convergent validity with other outcome measures. We included 42 pemphigus patients and 24 healthy individuals. A cross-sectional study was conducted in our Department of Dermatology at University of Pécs, Hungary. The EQ-5D was completed on the five-level version of EQ-5D (EQ-5D-SL) and Dermatology Life Quality Index (DLQI). Average gain at the time of the survey was evaluated by an 11-point visual analogue scale (VAS). Disease activity was measured by the Autimmune Bullous Skin Disorder Intensity Score (ABSIS). A total of 91 consecutive patients with pemphigus (pemphigus vulgaris: n = 69, pemphigus foliaceae: n = 29, other: n = 2) participated in the study. Median age was 57 years (range 19-93), and 69% were women. Overall, 47%, 44%, 43%, 40% and 17% of the patients reported problems regarding pain, mobility, anorexia, depression, usual activities and self-care, respectively. Median EQ-5D-SL index scores of patients with limited (n = 47), moderate (n = 22), severe (n = 17) and extensive (n = 4) pemphigus were as follows: 0.94, 0.86, 0.76 and 0.70 (p = 0.005). The EQ-5D-SL showed a strong correlation with the average pain VAS (r = -0.60; p < 0.001), and a moderate correlation with DLQI (r = -0.48; p = 0.001) and ABSIS scores (r = -0.41; p = 0.001). This is the first study in the literature to use the EQ-5D questionnaire in pemphigus. The EQ-5D is a valuable instrument for the assessment of HRQoL in pemphigus patients that can be also used to generate health utility values for economic evaluations of pemphigus treatments.

Psoriatic arthritis and hearing loss

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Psoriasis arthritis and hearing loss were examined in the first time. The impact of psoriatic arthritis on hearing impairment was investigated in this paper. Our aim is to investigate the association of hearing impairment and psoriasis arthritis, by using the Health Interview Survey (HIS). The hearing impairment and psoriasis arthritis are secondary aims. The purpose of this study is to investigate the impact of psoriatic arthritis on mental well-being. We used data from the National Health and Nutrition Examination Surveys for adults aged ≥20 years (n = 10,744). Association of psoriatic arthritis with above outcomes was modeled using multivariable generalized linear and ordinal logistic regression models, adjusted for demographics and medical comorbidities. Structural equation models (SEM) were developed to explore the extent to which hearing impairment mediates the effect of psoriatic arthritis on mental health. Psoriatic arthritis was present in 1.1% of the study population. Individuals with psoriatic arthritis were more likely to report hearing difficulties (OR 1.74, p = 0.005), seeing a mental health provider (OR 1.94, p = 0.019), have an average of 2.37 more days of poor mental health over last month (p = 0.002), and were more likely to be depressed (OR 2.68, p = 0.001), than normal controls. SEM analysis revealed that hearing impairment mediated 7.0%, 8.6%, and 6.4% of the effect of psoriatic arthritis on days of poor mental health, seeing a mental health provider, and depression, respectively. This study suggests that psoriatic arthritis is independently associated with a significantly increased risk of hearing impairment, which, in turn, partially mediates an association with poorer psychiatric outcomes. As such, the results of this study call for an increased awareness of these comorbidities when treating patients with psoriatic arthritis.

Clinical Outcomes | ABSTRACTS