Pigmentosa xeroderma group D: A clinical and genetic study of 19 Japanese cases
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Xeroderma pigmentosa (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 neurocutaneous disease, and XP/Cockayne syndrome (CS) complex. Genetically, XP is classified into two groups: one group classified by XPV and the other group classified by XPA. In Japan, a total of 257 cases were reported, which is the largest number worldwide. Among these, 89% of the cases were classified as XPA and 11% as XPV. In our study, we examined 91 Japanese XP cases, of which 76 were confirmed and 15 were suspected cases. Our results showed that there were 59 cases of XP patients with skin disease and 32 cases of XP patients with neurological disease. The most frequent skin disease was the appearance of skin lesions after sun exposure, and the most frequent neurological disease was the appearance of hearing loss, followed by spinal muscular atrophy. These results are consistent with previous reports on XP patients in Japan. However, there were some differences in the frequency of neurological disease between Japan and other countries. Therefore, our study suggests that further studies on the genetics and clinical features of XP patients in Japan are needed to better understand the disease in this population.

Apremilast in psoriasis - a prospective real-world study
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Apremilast is a novel oral phosphodiesterase-4 inhibitor approved for psoriasis treatment. Randomized trials have documented its efficacy and safety, but data on real-world patients are scarce. We aim to characterize psoriasis patients treated with apremilast in a real-world setting and calculate drug survival as an important measure of efficacy and compliance. All psoriasis patients that received apremilast between April 1st 2015 and January 19th 2017 were evaluated every 4 weeks and we documented: age, weight, height, smoking status, family history of psoriasis, joint involvement, previous treatments, psoriasis area severity index (PASI) scores, and the onset and duration of adverse events (AE). Efficacy was analyzed by PASI50, PASI75, and PAS90, reflecting the improvement of skin lesions compared to the PASI baseline. Kaplan-Meier statistics were used for drug survival estimates. Forty-eight patients were included. The median apremilast drug survival was 12.5 weeks (range 1–87). Three patients reached PASI75, four reached PASI90, and eight patients had PASI50. None of the obese patients (BMI>30, n=6) reached PASI75, compared to 12% of the non-obese patients (BMI<30, n=31). Thirty-one patients (64.6%) reported at least one AE, most frequently diarhoea (n=21, 43.8%), headache (n=7, 14.6%) and joint pain (n=5, 10.4%). We conclude that despite differences between real-world and trial patients, apremilast is safe and effective for the treatment of skin psoriasis in the daily practice. Up to 40% of patients will reach PASI50 or higher, but only few patients will reach PASI90.