◇Case Report

〈本文p.773～776〉

A three-generation Japanese family with punctate palmoplantar keratoderma type 1 caused by an *AAGAB* frameshift mutation, c.195_198de l4 (p.Lys66Phefs*42)

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Abstract
A 55-year-old female had suffered from multiple tiny hyperkeratotic lesions on her palms and soles for 20 years. Histopathology of the papules sampled from the left palm and sole showed hyperkeratosis with a grown granular layer. Her mother and son were also similarly affected. Direct sequencing identified a heterozygous 4-base pair deletion in exon 2 of *AAGAB*, c.195_198del4, resulting in a premature-termination codon (p.Lys66Phefs*42) within her, her mother, and her first-born son. Taken together, they were diagnosed with punctate palmoplantar keratoderma type 1. Oral etretinate was effective for the lesions.

〈本文p.777～780〉

A family of Cowden disease diagnosed by acquired lymphangiomas

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Abstract
We present a case of 38-year-old female of Cowden disease with vascular malformations. She complained acquired lymphangiomas on her left mamma, and she did not notice the characteristic lesions of Cowden disease. Because mucocutaneous lesions are occasionally mild, we should pay attention to their lesions. Vascular malformations have been rarely reported in Cowden disease, but we need to recognize them as clinical features of Cowden disease.
We performed genetic tests in her family after obtaining informed consent for the purpose of early detection of malignant lesions.

〈本文p.781～784〉

Compound heterozygous *TGM1* gene mutations in two male siblings with lamellar ichthyosis

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Abstract
We present two brothers affected by lamellar ichthyosis. Arcuate-shaped, large, brownish scales covered his face with ectropion. His lower legs were also covered with larger thick, brownish, plate-like scales. Electron microscopy showed that the marginal band was almost absent and *in situ* transglutaminase 1 activity was also absent in skin from the patient. We conclude that the severe phenotype of the patients developed due to the compound heterozygous null truncation mutations in *TGM1*.

◇Case Report

〈本文p.785～788〉

Three cases of Nagashima-type palmoplantar keratoderma with a mutation in *SERPINB7*

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Abstract
Case 1：A 42-year-old female. Case 2：A 36-year-old female. Case 3：A 1-year-old girl. Three patients were observed reddish hyperkeratosis of hands and feet extending to the dorsal areas of the fingers and toes after birth. We found *SERPINB7* mutations in these cases. Based on these results and the patients' clinical appearance, a diagnosis of Nagashima-type palmoplantar keratoderma were made in our cases.

〈本文p.789～792〉

Two familial cases of Birt-Hogg-Dubé syndrome

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Abstract
We described two familial cases with BHD syndrome, the probands of which presented with repetitive pneumothorax since their thirties. Both cases had multiple fibrofolliculomas and was diagnosed by skin biopsy and genetic testing. The skin lesions usually occur around the age of 25 and increase with age. Many cases only with skin symptoms might have been overlooked before the onset pneumothorax. Dermatologists have a role to suspect BHD syndrome from the skin manifestation for early definitive diagnosis, which requires a skin biopsy or genetic testing, before the occurrence of pneumothorax or renal tumor.

〈本文p.793～796〉

A case of Hailey-Hailey disease with a novel mutation in *ATP2C1* gene

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Abstract
A 51-year-old male had wettable erythema mainly in the axilla and groin since 2000. Histopathologically, the horny layer was thickened and the epidermis was separated at the suprabasal and middle layers, which was showing "dilapidated brick wall" appearance. Based on these findings, we diagnosed this case as Hailey-Hailey disease. In the genetic analysis, we identified a novel heterozygous c.234delG mutantion in exon3 of the *ATP2C1* gene.

◇Case Report

〈本文p.797～800〉

A case of Dowling-Meara type epidermolysis bullosa simplex

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Abstract
A 2-year-old girl was referred to our hospital due to blisters and erosions on her trunk and extrimities since at birth. Her family had no history of blistering disease.
Immunofluorescent study of the patient skin showed no apparent decreasing or defect of molecules expressed in basement membrane zone. Electron microscopy showed blister formation in the basal cells and aggregation of tonofilaments in the epidermal keratinocytes. Based on these findings, we diagnosed this case with Dowling-Meara type epidermolysis bullosa simplex.
The patient had substitution mutation (c.373C>T), which resulted in p.Arg125Cys in the *keratin 14* gene.

〈本文p.801～804〉

A case of KID syndrome accompanied by recalcitrant hyperkeratotic plaques on the lower legs

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Abstract
A 20-year old man formerly genetically diagnosed with keratitis ichthyosis deafness (KID) syndrome had been suffering from recalcitrant bilateral lower leg hyperkeratotic plaques. Histologically the lesion showed pseudoepitheliomatous hyperplasia with granulation tissue formation and candidial hyphal components at the horny layer. Oral and topical antifungal agent administration reduced the area of keratinization and granulation. It was speculated by literature review that the unique hyperkeratotic lesion with candidial or other fungal infection is one of the 'extra-triad' of KID syndrome and this usually requires long-term antifungal treatment.

〈本文p.805～808〉

A case of vascular-type Ehlers-Danlos syndrome

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Abstract
A 36-year-old man developed compartment syndrome to his right calf without triggers like exercising or injury. At an operation, we found the posterior tibial artery ruptured. His skin was thin and joints of hands and fingers were extended excessively. From the analysis of the level of synthesized collagens by cultured dermal fibroblasts, the amount of synthesized type Ⅲ collagen was significantly reduced in the fibroblasts from the patient. From the DNA sequence analysis of type Ⅲ collagen (COL3A1), a heterozygous point mutation of COL3A1 that resulted in a glycine substitution was shown. We diagnosed him as vascular type of Ehlers-Danlos syndrome and started to medicine him celiprolol.

◇Case Report

〈本文p.809～812〉

A case of Werner syndrome

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Abstract
A 45-year-old male was complained of intractable skin ulcer of left leg. He was short-statured and lighter build. His face was bird-like and his hair was gray and bold. His voice was high pitched and squeaky. A genetic testing revealed he was homozygous of Type4 mutation of *WRN*. Pathognomonical signs and the mutation of *WRN* provided a definite diagnosis of Werner's syndrome.

〈本文p.813～816〉

Case of pachydermoperiostosis with compound heterozygous mutations in the *SLCO2A1* gene

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Abstract
A 26-year-old man was referred to us because of a 10-year history of digital clubbing, progressive thickening and furrowing of forehead and scalp skin, oily skin, acne, palmoplantar excoriation due to hyperhidrosis, associated with arthralgia and a low grade fever. The patient was diagnosed as having pachydermoperiostosis from the typical clinical features and periosteal thickening of the long bones by radiological examination. The patient had no family history of pachydermoperiostosis. Sanger sequencing identified heterozygous mutations including c.940+1G>A (intron 7) and c.664G>A (exon 5) in the *SLCO2A1* gene.

〈本文p.817～820〉

Xeroderma pigmentosum variant type who has not developed skin cancer

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Abstract
A 28-year-old female suggestive of xeroderma pigmentosum (XP) came to our department 10 years before. At that time she was strongly suggestive of XP variant type (XP-V) because *POLH* mutations were identified in one allele. Because patients with XP-V do not manifest abnormally exaggerated sunburn, they are often diagnosed at the occasion of their consultation of skin cancers. In our case, owing to early diagnosis and protection from light, no skin cancer has developed.

◇Case Report

〈本文p.821～824〉

Two cases of autosomal recessive woolly hair with *LIPH* gene mutation

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Abstract

We describe two cases of autosomal recessive woolly hair. The first case was a 36-year-old woman with woolly and sparse hairs on her scalp without other abnormalities. The mutation analysis revealed that the patient has a homozygous missence mutation c.736T＞A in exon6 of the *LIPH* gene. The second case was a 41-year-old woman with woolly hair without other abnormalities. She has a heterozygous missense mutation c.736T＞A and c.742C>A in exon6 of the *LIPH* gene.

On the basis of clinical and genetic findings, we diagnosed these cases as autosomal recessive woolly hair by *LIPH* gene mutation.

〈本文p.825～828〉

A case of clear cell sarcoma of the finger

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Abstract

A 37-year-old female presented with a mass lesion on the middle finger of her right hand of one year and half duration. Biopsy of the mass lesion showed abundant nests of round cells with clear cytoplasm. On immunohistochemical examination, tumor cells were immunoreactive for HMB-45 and Melan A. FISH (fluorescence *in situ* hybridization) analysis showed reconstitution of *EWSR1* (Ewing sarcoma breakpoint region 1). We diagnosed this tumor as clear cell sarcoma.

〈本文p.829～832〉

A case of subcutaneous panniculitis-like T-cell lymphoma in a senior

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Abstract

A 80 year-old woman presented with a lesion of 10-year history of her right leg. Upon a physical examination, brown purplish colored induration sized 12.5×13 cm could be palpated on the right lower leg. Histopathologically, basophilic mitotic cells hyperplasia forming rimmig was found at the area from upper dermis to fatty tissue. Immunohistochemical examination showed positive staining for CD3 and CD8, but negative staining for CD4 and CD56. T-cell receptor gene reconstruction was found at Cβ1 area and not found in Jδ1area. Based on these findings, she was diagnosed as subcutaneous panniculitis-like T-cell lymphoma. THP-COP was effective in this case.