Facial angiofibromas are main characteristics of tuberous sclerosis complex (TSC), an autosomal dominant genodermatosis. Treatment of angiofibromas has been limited to surgically destructive techniques, including ablative laser therapy. Recently, the clinical trials have revealed that topical rapamycin (sirolimus) has been shown to be safe and beneficial effect of the facial angiofibroma. We herein report on a 23-year-old woman who treated successfully by a combination of fractional resurfacing and topical rapamycin.

Keyword: Facial angiofibroma, Topical Rapamycin

P285

Two cases of mild piebaldism with novel mutations in the tyrosine kinase domain of KIT gene

Department of Dermatology, Severance Hospital, Cutaneous Biology Research Institute, Yonsei University College of Medicine, 1Department of Clinical Genetics, Yonsei University College of Medicine, 2Department of Dermatology, Yamagata University School of Medicine, Yamagata, Japan

Hemin Lee1, Sang Ho Oh1, Kyo-Yeon Koo1, Tamio Suzuki2, Jin-Sung Lee1

Piebaldism is a rare dermatologic disorder inherited as an autosomal trait. Mutation of the KIT proto-oncogene encoding transmembrane receptor tyrosine kinase (TK) is responsible for the disease, and its clinical manifestations and phenotypic severity may depend on the mutation site. We report two cases of mild piebaldism in Korean patients associated with mutations in TK domain of the KIT gene. Although mutations in highly conserved cytoplasmic TK region were previously known to be associated with severe phenotypes, clinically milder forms can be presented with other functioning modifier genes such as MC1R or polymorphism in the extracellular domain. More exhaustive studies on KIT gene and other modifier factors are needed to reveal new insights into various phenotypes of piebaldism and melanogenesis.

Keyword: Piebaldism, KIT, Tyrosine kinase, TK

P286

A case of netherton syndrome responsive to topical pimecrolimus

Department of Dermatology & Cutaneous Biology Research Institute, Yonsei University College of Medicine, Seoul, Korea

Hemin Lee, Jungsoo Lee, Jung U Shin, Kwang Hoon Lee

Netherton syndrome is a rare autosomal recessive disorder with a triad of ichthyosis, hair shaft abnormality, and atopic diathesis. We report a case of a 17-year-old male who had bright red face and brittle hair since his birth and developed polycyclic serpiginous lesions and peripheral scales on extremities during youth. Clinically the lesions were compatible with ichthyosis linearis circumflexa (ILC). Atopic skin features were also prominent with elevated serum IgE and specific antibody titer to house dust mites. Trichorrhexis invaginata was identified through light and scanning electron microscopic examination. Diagnosed as Netherton syndrome, the patient applied topical pimecrolimus 1% ointment once daily for 6 months. No adverse events occurred, and the patient showed reduction in scaling and pruritus. However, facial erythema remained. Although pimecrolimus is effective and well-tolerated agent for ILC and atopic dermatitis, more attempts should be tried for improving facial erythema and hair fragility.

Keyword: Netherton syndrome, Ichthyosis linearis circumflexa, Trichorrhexis invaginata, Atopic dermatitis, Pimecrolimus

P287

A case of multiple, recurrent pilomatricoma in association with sex chromosomal abnormality, 47+XXY

Department of Dermatology, Chonnam National University Medical School, Gwangju, Korea

Ho-June Lee, Sook Jung Yun, Je-Bum Lee, Seong-Jin Kim, Seung-Chul Lee, Young Ho Won

Pilomatricoma, also known as Malherbe calcifying epithelioma and pilomatrixoma, is a benign neoplasm which is derived from hair matrix cells. It usually presents as a solitary, skin-colored or bluish, firm, cystic nodule on the head, neck, or proximal upper extremities. Multiple lesions are seen in Gardner's syndrome, myotonic dystrophy, Rubinstein-Taybi syndrome. Some cases have been reported to be associated with sarcoidosis, HIV, Soto syndrome, and chromosomal abnormality like Turner
syndrome and trisomy 9. A 4-year-old girl presented with multiple variable sized, skin colored subcutaneous nodules on back and right calf which were sequentially developed and removed surgically for six months. They all were diagnosed as pilomatricomas by the histopathological examination. In infancy, she had a developmental disability. For further evaluation, Brain MRI was checked and an arachnoid cyst was also detected. The chromosomal study reveals sex chromosomal abnormality, female type of 47+XXY. She also had an eyeball deviation and a lacrimal duct disorder. Four years later, pilomatricoma-like nodules were presented on left lower leg and left thigh, which diagnosed as pilomatricoma again. It should be rare that multiple, recurrent pilomatricoma and other symptoms described above may be related with sex chromosomal abnormality 44+XXY, and herein we report this unusual case.

Keyword: Pilomatricoma, Sex chromosomal abnormality, 47+XXY

P288

MIDAS syndrome diagnosed as linear skin atrophy on face
Department of Dermatology, School of Medicine, Chungnam National University, Daejeon, Korea
Seul-Ki Lim, Young Lee, Young-Joon Seo, Jeung-Hoon Lee, Myung Im

MIDAS syndrome is an X-linked dominant disease which is acronym of microphthalmia, dermal aplasia, and sclerocornea. In most patients, the unbalanced translocation or deletion of X chromosome short arm 22.3 band is observed. This disease characteristically shows linear atrophy of the skin limited to the face and neck, and congenital eye disease is accompanied. A 9-month-old female who has linear skin atrophy on right chin visited our clinic. She also presented microphthalmia and sclerocornea on her right eye. On chromosomal study, there was a deletion of X chromosome short arm 22.31 band. Here, we report the MIDAS syndrome patient who has linear skin atrophy on face.

Keyword: Segmental neurofibromatosis, Visceral neurofibroma, Mesentery, Omentum

P290

A case of Waardenburg syndrome with characteristic clinical features
Department of Dermatology, Samsung Medical Center, Sungkyunkwan University

Waardenburg syndrome is a rare inherited condition characterized by sensory/neural hearing loss, pigmentary abnormalities of the skin, hair, and eyes, and craniofacial anomalies and is classified as a disorder of neural crest