EDITORIAL

Management of autoimmune urticaria
Arun C. Inamadar, Aparna Palit ................................................................. 89

VIEWPOINT

Cosmetic dermatology versus cosmetology: A misnomer in need of urgent correction
Shyam B. Verma, Zoe D. Draelos ........................................................................ 92

REVIEW ARTICLE

Psoriasiform dermatoses
Virendra N. Sehgal, Sunil Dogra, Govind Srivastava, Ashok K. Aggarwal .......... 94

ORIGINAL ARTICLES

A study of allergen-specific IgE antibodies in Indian patients of atopic dermatitis
V. K. Somani ....................................................................................................... 100

Chronic idiopathic urticaria: Comparison of clinical features with positive autologous serum skin test
George Mamatha, C. Balachandran, Prabhu Smitha ........................................... 105

Autologous serum therapy in chronic urticaria: Old wine in a new bottle
A. K. Bajaj, Abir Saraswat, Amitabh Upadhyay, Rajetha Damisetty, Sandipan Dhar ........................................................................ 109

Use of patch testing for identifying allergen causing chronic urticaria
Ashimav Deb Sharma .......................................................................................... 114

Vitiligoid lichen sclerosus: A reappraisal
Venkat Ratnam Attili, Sasi Kiran Attili ............................................................... 118
**BRIEF REPORTS**

Activated charcoal and baking soda to reduce odor associated with extensive blistering disorders  
Arun Chakravarthi, C. R. Srinivas, Anil C. Mathew .......................................................... 122

Nevus of Ota: A series of 15 cases  
Shanmuga Sekar, Maria Kuruvila, Harsha S. Pai ............................................................................. 125

Premature ovarian failure due to cyclophosphamide: A report of four cases in dermatology practice  
Vikrant A. Saoji .................................................................................................................. 128

**CASE REPORTS**

Hand, foot and mouth disease in Nagpur  
Vikrant A. Saoji .................................................................................................................. 133

Non-familial multiple keratoacanthomas in a 70 year-old long-term non-progressor HIV-seropositive man  

Late onset isotretinoin resistant acne conglobata in a patient with acromegaly  
Kapil Jain, V. K. Jain, Kamal Aggarwal, Anu Bansal .............................................................. 139

Familial dyskeratotic comedones  
M. Sendhil Kumaran, Divya Appachu, Elizabeth Jayaseelan ..................................................... 142
Nasal NK/T cell lymphoma presenting as a lethal midline granuloma
Vandana Mehta, C. Balachandran, Sudha Bhat, V. Geetha, Donald Fernandes 145

Childhood sclerodermatomyositis with generalized morphea
Girishkumar R. Ambade, Rachita S. Dhurat, Nitin Lade, Hemangi R. Jerajani 148

Subcutaneous panniculitis-like T-cell cutaneous lymphoma
Avninder Singh, Joginder Kumar, Sujala Kapur, V. Ramesh 151

LETTERS TO EDITOR

Using a submersible pump to clean large areas of the body with antiseptics
C. R. Srinivas 154

Peutz-Jeghers syndrome with prominent palmoplantar pigmentation

Stratum corneum findings as clues to histological diagnosis of pityriasis lichenoides chronica
Rajiv Joshi 156

Author’s reply
S. Pradeep Nair 157

Omalizumab in severe chronic urticaria
K. V. Godse 157

Hypothesis: The potential utility of topical eflornithine against cutaneous leishmaniasis
M. R. Namazi 158

Nodular melanoma in a skin graft site scar
A. Gnaneshwar Rao, Kamal K. Jhamnani, Chandana Konda 159
<table>
<thead>
<tr>
<th>Title</th>
<th>Authors</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Palatal involvement in lepromatous leprosy</td>
<td>A. Gnaneshwar Rao, Chandana Konda, Kamal Jhamnani</td>
<td>161</td>
</tr>
<tr>
<td>Unilateral nevoid telangiectasia with no estrogen and progesterone receptors in a pediatric patient</td>
<td>F. Sule Afsar, Ragip Ortac, Gulden Diniz</td>
<td>163</td>
</tr>
<tr>
<td>Eruptive lichen planus in a child with celiac disease</td>
<td>Dipankar De, Amrinder J. Kanwar</td>
<td>164</td>
</tr>
<tr>
<td>Xerosis and pityriasis alba-like changes associated with zonisamide</td>
<td>Feroze Kaliyadan, Jayasree Manoj, S. Venkitakrishnan</td>
<td>165</td>
</tr>
<tr>
<td>Treatment of actinomycetoma with combination of rifampicin and co-trimoxazole</td>
<td>Rajiv Joshi</td>
<td>166</td>
</tr>
<tr>
<td>Vitiligo, psoriasis and imiquimod: Fitting all into the same pathway</td>
<td>Bell Raj Eapen</td>
<td>169</td>
</tr>
<tr>
<td>Author’s reply</td>
<td>Engin Şenel, Deniz Seçkin</td>
<td>169</td>
</tr>
<tr>
<td>Multiple dermatofibromas on face treated with carbon dioxide laser: The importance of laser parameters</td>
<td>Kabir Sardana, Vijay K. Garg</td>
<td>170</td>
</tr>
<tr>
<td>Author’s reply</td>
<td>D. S. Krupa Shankar, A. Kushalappa, K. S. Uma, Anjay A. Pai</td>
<td>170</td>
</tr>
<tr>
<td>Alopecia areata progressing to totalis/universalis in non-insulin dependent diabetes mellitus (type II): Failure of dexamethasone-cyclophosphamide pulse therapy</td>
<td>Virendra N. Sehgal, Sambit N. Bhattacharya, Sonal Sharma, Govind Srivastava, Ashok K. Aggarwal</td>
<td>171</td>
</tr>
<tr>
<td>Subungual exostosis</td>
<td>Kamal Aggarwal, Sanjeev Gupta, Vijay Kumar Jain, Amit Mital, Sunita Gupta</td>
<td>173</td>
</tr>
</tbody>
</table>
Clinicohistopathological correlation of leprosy
Amrish N. Pandya, Hemali J. Tailor ................................................................. 174

RESIDENT’S PAGE

Dermatographism
Dipti Bhute, Bhavana Doshi, Sushil Pande, Sunanda Mahajan, Vidya Kharkar ................................................................. 177

FOCUS

Mycophenolate mofetil
Amar Surjushe, D. G. Saple ........................................................................ 180

QUIZ

Multiple papules on the vulva
G. Raghu Rama Rao, R. Radha Rani, A. Amareswar, P. V. Krishnam
Raju, P. Raja Kumari, Y. Hari Kishan Kumar .................................................. 185

Net Study
Oral isotretinoin is as effective as a combination of oral isotretinoin and topical anti-acne agents in nodulocystic acne
Rajeev Dhir, Neetu P. Gehi, Reetu Agarwal, Yuvaraj E. More ................................................................. 187

Net Case
Cutaneous diphtheria masquerading as a sexually transmitted disease
T. P. Vetrichevvel, Gajanan A. Pise, Kishan Kumar Agrawal,
Devinder Mohan Thappa ........................................................................ 187

Net Letters
Patch test in Behcet’s disease
Ülker Gül, Müzeyyen Gönül, Seray Külüç Çakmak, Arzu Kılıç ................................................................. 187

Cerebriform elephantiasis of the vulva following tuberculous lymphadenitis
Surajit Nayak, Basanti Acharjiya, Basanti Devi, Satyadarshi Pattnaik,
Manoj Kumar Patra ........................................................................ 188

Net Quiz
Vesicles on the tongue
Saurabh Agarwal, Krishna Gopal, Binay Kumar .................................................. 188

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Letters to Editor

Peutz-Jeghers syndrome with prominent palmoplantar pigmentation

Sir,

Peutz-Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by lentigines on the lips, buccal mucosa, tips of fingers and toes in association with intestinal polyps. PJS is caused by mutations in the gene coding for serine threonine kinase, located on the p arm of chromosome 19.[1,2] The pigmented macules that usually appear during infancy and early childhood, are seen over the back of the hands and tips of the fingers and toes but are very rarely seen over the palms and soles.[3]

A 17-year-old male was referred from the surgery department to us for evaluation of the pigmented lesions over his palms and soles. Recently, he was operated for persistent colicky abdominal pain and was found to have an exophytic growth in the jejunum, which showed evidence of villous adenomas on histopathological examination. On enquiry, the patient gave a history of pigmented lesions over the palms and soles as well as the lips since childhood that increased in size and number over the past year. Two of his siblings also
had history of pigmentation of the lips as well as their palms and soles. On examination, multiple discrete-to-coalescent, hyperpigmented macules 0.5-1.5 mm in size were seen over the lips [Figure 1], perioral area and buccal mucosa. Similar hyperpigmented and round-to-oval macules of varying sizes ranging from 3 mm to more than 1 cm in diameter were present over the tips of the fingers and up to 5 cm in diameter were seen over the soles [Figure 2]. A 12 cm-long paramedian linear scar was evident over the abdomen. A diagnosis of Peutz-Jeghers syndrome was made.

The pigmented macules of Peutz-Jeghers syndrome (PJS) usually appear during infancy and early childhood, but may be present at birth or develop later in life and have a tendency to increase in size during adolescence. Round, oval or irregular patches of brown or almost black pigmentation, 1-5 mm in diameter, are irregularly distributed over the buccal mucosa, gums, hard palate and lips, especially the lower lip. The pigmented macules may also occur on the face especially around the nose and the mouth but are smaller and often under 1 mm in size. Large macules are rarely seen over the back of the hands and the tips of the fingers and toes.[10] Our case had large hyperpigmented macules over both his palms and soles [Figure 2], which is very unusual.

Intestinal manifestations include numerous polyps in the jejunum, ileum and less frequently in the colon, rectum, stomach and duodenum. Patients with PJS have a 10-18 fold greater lifetime cancer risk than the general population. The greatest risk is for gastrointestinal malignancy of the colon and duodenum.[14] Laugier-Hunziker syndrome and Cronkhite-Canada syndrome should be considered in the differential diagnosis of PJS. In the former, mucosal and nail pigmentation are characteristic but without intestinal polyps, whereas the vular aspects of the fingers are heavily pigmented along with evidence of intestinal polyps but without mucosal pigmentation in the latter.[5,6]

Although perioral pigmentation is commonly seen in PJS, palmoplantar pigmentation is rare and such numerous large hyperpigmented macules over the soles have never been described before and hence, being reported.

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