Genodermatoses in the Gulf Countries

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Abstract
Genodermatoses in the Gulf countries have been reviewed. The high occurrence of diseases like epidermolysis bullosa, mal de Meleda and biotinidase deficiency have been noted. New syndromes are also continuously being reported. Molecular studies have revealed findings different from those described elsewhere. These have been linked to the large family size and tribal customs that lead to consanguinity. Influx of immigrants, wars and environmental pollutants are likely to worsen the situation. As treatment of genetic disorders is a challenging issue, prevention must be targeted by providing good premarital counseling and altering the tribal customs that promote consanguinity.

Introduction
Of the non-communicable diseases, genetic diseases are a major cause of both morbidity and mortality. There are isolated reports on a number of genetic conditions in the Arab population. These try to unveil the complexities of inheritance in a rapidly emerging population with common cultural, historical, traditional and religious ties. The large family size, high rate of consanguinity and other permitted forms of intermarriage make the Arabs an ideal group for the study of genetically inherited diseases. Most studies have concentrated on blood disorders, the commonest ones seen in the gulf. This prompted us to review the genodermatoses seen in the region.

Genodermatoses are inherited disorders determined by chromosomal aberrations or a single gene factor.1 As disorders due to single gene factors are common and affect many people, these dermatoses in the gulf have been reviewed to acquaint the reader with the prevailing situation. The tables list the genodermatoses reported in the states of the Gulf Coop-

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reported. An array of overlapping features have been seen in related syndromes namely, infantile systemic hyalinosis and juvenile systemic fibromatosis, gero-
dermia osteodysplastica and wrinkly skin syndrome, and reticulate acropigmentation of Kitamura and Dowling-Degos disease.

The high incidence of inherited defects also explains the appearance of new syndromes with cutaneous abnormalities in the gulf states. They have been seen as keratinization defects and hair anomalies, loose or lax skin and joints with other defects, or associated with known syndromes, and along with a variety of dysmorphic features. Some appear to be newer variants of Ehlers-Danlos syndrome while another one has been eponymously named Teebi overgrowth syndrome.

Limited studies in some conditions at the molecular level have revealed that the findings are at variance with those seen elsewhere. These include ataxia telangiectasia, ichthyosis, and auto-
somal recessive congenital ichthyosis and autosomal recessive hypotrichosis simplex. Unique mutations have been described in dyskeratosis congenita, junctional epidermolysis bullosa and mal de Meleda.

**Therapy:**

Treatment of genodermatoses can be challenging. Though some like lethal congenital erythroderma are not compatible with life, there are others where the quality of life can be improved by treatment. These are often metabolic disorders and range from mere reversal of hypopigmentation in homocystinuria with pyridoxine to dramatic response in Richner Hanhart syndrome to diet restriction in tyrosine and phenylalanine. Unrecognized, a condition like multiple carboxylase deficiency which responds to biotin can be fatal. The advent of retinoids has also reversed the prognosis of some genodermatoses: sebaceous gland hyperplasia and Papillon-Le fevre syndrome are good examples and an encouraging response has been seen in har-lequin ichthyosis. Advanced therapy like bone marrow transplantation has been done with success in Chediak-Higashi syndrome.

**Conclusion:**

It would be obvious from the reports that many patients are strictly not gulf citizens but hail from other countries like Sudan, Egypt, Syria, Turkey, Palestine and Pakistan. As the oil driven economy of the gulf states accelerates progress the influx would be more from other countries. Many stay for long or even settle. Wars too contribute to environmental changes that can lead to mutation. These factors are likely to add new diseases that may make their appearance later. It is difficult to intervene here but what is needed now is a programme for routinely screening the common genodermatoses and identifying those at risk. Prevention should be brought about by appropriate premarital counseling and discouraging consanguineous marriages. To effectively channelise these services to the community, a Center for Arab Genetic Studies was recently established in June 2003 in Al Wasl Hospital, Dubai, U.A.E. which can be reached at www.cags.org. ae.

**References**


77. Lestratinng AG, Hadi SM, Qayed KI, Diaynej B. Mal de Meleda: recessive transgressive palmoplantar keratoderma with three unusual facultative features. Dermatology 1992;184:78-82


Table 1: Genodermatoses in the Gulf

<table>
<thead>
<tr>
<th>S.No</th>
<th>Disorder/Phenotype</th>
<th>No. of patient(s) &amp; Nationality</th>
<th>OMIM No.</th>
<th>Inheritance*</th>
<th>Clinical Cutaneous</th>
<th>Features*Other anomalies/Systems affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>APPENDAGEAL LOSS</td>
<td>1. Anhidrotic ectodermal dysplasia **</td>
<td>6 Qatar, Sudan, Palestine</td>
<td>129490 224900</td>
<td>AD or AR**</td>
<td>Absent or diminished sweat &amp; hypotrichosis</td>
</tr>
<tr>
<td>1</td>
<td>2. Anonychia congenita simplex **</td>
<td>1</td>
<td>206800</td>
<td>AD, AR or sporadic</td>
<td>Absence of nails</td>
<td>None</td>
</tr>
<tr>
<td>2</td>
<td>3. Atrichia congenita **</td>
<td>3 Saudi Many Oman</td>
<td>203655</td>
<td>AR</td>
<td>Total absence of hairs Keratin cysts on face &amp; limbs</td>
<td>None</td>
</tr>
<tr>
<td>3</td>
<td>4. Chondroectodermal dysplasia (Ellis-van Creveld syndrome) **</td>
<td>3 Yemen and Saudi families</td>
<td>209500</td>
<td>AR</td>
<td>Hypoplastic nails</td>
<td>Dwarfism, Teeth, Cardiac defects in 50%</td>
</tr>
<tr>
<td>4</td>
<td>5. Frontofacionasal dysplasia **</td>
<td>1UAE **</td>
<td>225500</td>
<td>AR</td>
<td>Hypoplasia of multiple skin appendages</td>
<td>Multiple facial defects, CNS</td>
</tr>
<tr>
<td>5</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>Hereditary hypotrichosis simplex</td>
<td>4</td>
<td>Saudi</td>
<td>229400 146520</td>
<td>AD</td>
<td>Progressive hair loss leading to baldness by age of 30</td>
</tr>
<tr>
<td>7</td>
<td>Leuconychia totalis</td>
<td>1</td>
<td>UAE</td>
<td>151600</td>
<td>AD, AR</td>
<td>White nails, Hyperhidrosis</td>
</tr>
<tr>
<td>8</td>
<td>Primary hypogonadism and partial alopecia</td>
<td>3</td>
<td>Kuwait</td>
<td>Not available</td>
<td>AR</td>
<td>Alopecia</td>
</tr>
<tr>
<td>9</td>
<td>Vitamin D resistant rickets type II with alopecia</td>
<td>4</td>
<td>Kuwait/Saudi</td>
<td>Not listed</td>
<td>AR</td>
<td>Alopecia</td>
</tr>
<tr>
<td>10</td>
<td>Woodhouse &amp; Sakati syndrome</td>
<td>6</td>
<td>Saudi</td>
<td>241080</td>
<td>AR</td>
<td>Alopecia</td>
</tr>
</tbody>
</table>

**B**

**PIGMENTARY CHANGES**

| 1 | Chediak-Higashi syndrome | 7 | Saudi | 214500 | AR | Albinism | Eyes |
| 2 | Dyschromatosis universalis hereditaria | 1 | Saudi | 127500 | AR | Leukomela noderm | None |
| 3 | Fanconi's anemia | 1 | Saudi | 227650 | AR | Pigmentary changes | Bone marrow, Heart, Kidneys, Malformed limbs |
| 4 | Incontinentia pigmenti | 4 | Qatar/Saudi | 308300 | X-linked dominant | Typical hyperpigmentation | CNS, Skeletal, Eyes, Teeth |
| 5 | Peutz-Jegher's syndrome | 1 | Saudi | 175200 | AD | Acral melanosis | Gastrointestinal tract |
| 6 | Piebaldism | 3 | Qatar | 17280 | AD | White patches on skin and scalp | None |
| 7 | Reticulate pigmentation of Dohi | 3 (AR) | | 127400 | AD, rarely AR | Acral localized pigmentation | None |
| 8 | Reticulate acropigmentation of Kitamura | 1 | UAE | Not given | AD | Acral melanosis, palmar pits & no hypo pigmentation | None |

**C**

**ICHTHYOTIC SKIN**

| 1 | Autosomal recessive congenital ichthyosis (ARCI) | 2 | UAE families | 242100 | AR | Variable erythema & scaling | None |
| 2 | Chanarin-Dorfman syndrome (lipid storage disease) | 2 | Saudi | 275630 | AR | Congenital ichthyotic erythroderma | Multiple systems |