Tel-Hashomer Camptodactyly Syndrome with Hirsuitism in an Indian Family

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Abstract
Two new cases with the Tel-Hashomer camptodactyly syndrome have been ascertained in an Indian family. This report emphasizes the autosomal recessive nature of disease and documents an additional feature of hirsuitism not previously described. The gene for Tel-Hashomer camptodactyly syndrome is present in all populations around the world.

INTRODUCTION
Goodman et al described the Tel-Hashomer syndrome in two sibling born to non-consanguineous parents. He further observed two additional cases with the syndrome. Gollop and Colleto described patients from two Brazilian families having consanguinity. Patton et al in showed that muscle weakness was due to abnormal muscle histology. Tylki-Szymanska reported two cases of the syndrome in one Arabic family, both born to first cousins. Pagnan et al described two siblings from a Brazilian family. Toriello described two siblings with mitral valve prolapse in a Hispanic boy and girl. To date around 30 cases have been reported around world wide.

We describe two sisters from a non-consanguinous Indian family with all features of Tel-Hashomer syndrome as well as hirsuitism a feature not previously described.

CASE REPORT

Patient 1
The propositus is a 30 year old Indian girl who was born at term. Birth weight was 2.4 kg. She was born to non-consanguineous Indian parents.

On physical examination, height, weight and circumference of head was (50th%tile). She had down-slanting palpebral fissures, hypertelorism, high arched palate, webbing of neck, narrow shoulders, low-set ears and attached lobule, small mouth, maloccluded teeth, camptodactyly, single palmar crease, flat feet and marked hirsuitism. Intellectual development was normal. Cytogenetic analysis of peripheral blood showed a 46, XX, chromosomal complement. Hormonal profiles were within normal limits. Estimation of dihydroepiandrosterone was mildly elevated. Ultrasonography of pelvis and adrenals were normal.

Patient 2
Her younger sister was born after an uneventful gestation. On physical examination. She had similar facial features and short stature, camptodactyly abnormal dermatographics and normal intelligence. There was no history of any other family members affected except the elder sister. An unusual feature was hirsuitism. Cytogenetic analysis of peripheral blood revealed 46, XX, chromosomal complement.
DISCUSSION

All patients described up to now show striking clinical similarity. All have facial asymmetry, small mouth, maloccluded teeth, downslanting palpebral fissures, camptodactyly and muscle hypotonia. Frequent findings are brachycephaly, crowded teeth, thoracic scoliosis, cardiovascular disorders, nervous system and muscle disorders. Both our patients had all features of Tel-Hashomer Syndrome. An unusual finding was hirsuitism. This has not been documented before. Mildly elevated levels of DHEA is suggestive of adrenal steroidogenesis.

Autosomal recessive inheritance is confirmed in this report.

Since Tel-Hashomer syndrome is reported in Sepahardic Jews in Israel, Moslems in Libya, Anglo-Asians, Brazilians and Indians it suggests that the gene pool is present in all groups of population worldwide.

REFERENCES


Announcement

HIV Congress 2005

11th to 13th March, 2005 (Jointly organized by Indian Academy of HIV and Liver Diseases, AIDS Society of India, Jaslok Hospital and Research Centre, and Bhatia Hospital and Research Centre).

Venue: The Taj President Hotel, Mumbai

Pre-conference workshop: Clinical presentation of interesting HIV patients and few of plenary sessions on 11th March 2005. SP Jain Auditorium, Bombay Hospital and Research Centre.

Congress Secretariat: Dr. JK Maniar, Organising Chairperson, Email: jkmaniar@vsnl.com; Fax: 91-22-22083184

Announcement

The Department of Tuberculosis and Chest Diseases is organizing a "National Seminar on Tuberculosis and Chest Diseases" at Institute of Medical Sciences, Banaras Hindu University, Varanasi, 221 005 (UP) on February 20, 2005 (Sunday).

For further information contact: Dr. JK Mishra, Organising Secretary, National Seminar on TB and Chest Diseases, Room No. 1352, Department of TB and Respiratory Diseases, Institute of Medical Sciences, Banaras Hindu University, Varanasi 221 005 (UP).
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