Original Article

Familial Ectrodactyly and Its Ocular Associations

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Purpose: To report a novel family of Ectrodactyly and its associations with eye pathology.

Material And Methods: This observational and analytical study was conducted at Allied Hospital, Punjab Medical College (PMC), Faisalabad, from March 15 to April 15, 2007. A detailed history, examination and investigation of three families of Ectrodactyly were completed.

Results: All members of three families had same findings in hands and feet. All were mentally sharp. They were all suffering from Myopia and cataract.

Conclusion: The familial Ectrodactyly is associated with Myopia and cataract. No other eye findings were seen.

The word "Ectrodactyly" is derived from the Greek "Ektrona" (abortion) and "Dactylos" (Finger) literally "Abortion of a finger". The Ectrodactyly, commonly known as "lobster claw syndrome"[1], sometimes known as "Karsch-Neugebauer syndrome", is a rare congenital deformity of the hand where the middle digit is missing and the hand is cleft. Ectrodactyly is an inherited dysmelia, and often occurs in both the hands and the feet. Its inheritance pattern is autosomal dominant. It affects about 1 in 90,000 babies, with males and females equally are affected. The type I, the most frequent form has been found to be a mutation on chromosome 7 in a region that contains two homeobox genes, DLX5 and DLX6[2].

The associated eye findings are seen in many systemic diseases and also in familial ectrodactyly but these are uncommon and rare. Some of these associated ocular findings have already been reported but our associated findings (myopia and cataract) have never been reported in the literature earlier.

MATERIAL AND METHODS

Three families with Ectrodactyly were evaluated and detailed history of birth, medical, surgical and family history was taken including history of drugs, irradiation and maternal ailment. Examination included visual acuity, refraction, slit lamp, posterior segment examination, tonometry and gonioscopy was performed. The investigations e.g. Blood complete examination, Urine complete examination, ESR, Blood Sugar random, Growth hormones estimation, RA factors, ANA, VDRL, Urine for reducing substances, RBC Hexokinase enzyme, urine chromatography and Calcium and Phosphorus determination was done.

RESULTS

In all the members of three families had the same findings of hands and feet. All the members were healthy and mentally sharp. In family no. 1, one affected father having unaffected wife had five children (M4 + F1). All the five children had hands abnormalities (100 %). Out of five, 2 (40 %) had feet and eye findings (myopia + lens changes), while 3(60%) were unaffected. In family no. 2, affected female having unaffected husband was bearing four

children. All of them had hands and feet abnormalities (100 %). Her one affected male child died at the age of five years. Out of living four, 2 (50 %) had eye findings (myopia + lens changes) and 2 (50 %) were unaffected. The third family comprised of affected mother and normal husband. This family also had four children. All had hands and feet abnormalities, but 2 (50 %) had eye findings (myopia + lens changes) and other 2 (50 %) were normal (Table 1-4). We examined three families. These were close relatives and had close and inter marriage system (marriages between two families on exchange basis). The other pedigree shown in diagram was made by history. They narrated that problem started after a great grandmother who had hands and feet abnormalities. Close inter marriage system increased the frequency of occurrence of pathology. None of the patients had other eye pathologies except lens changes and myopia. Also, none of the patients had uveitis, glaucoma, ectopia lentis, and retinal detachment. No patient had marfanoid features, blond hair, mental retardation, hirsutism, moon face, mouth or feet ulcers, pathological fractures, serositis, hepatomegaly, lymphadenopathy etc.

DISCUSSION

Ectrodactyly is a congenital defect that causes malformation of the hands alone or with feet. In this condition, the middle finger or middle toe is missing. Currently there are several treatments, which can normalize the appearance of the hands, yet they will not function precisely the same way as regularly formed hands. Some people with Ectrodactyly use prosthetic hands to avoid the rude stares of others. The "Upton" a micro surgeon, said that toes are spare parts available for transfer. He used the children's tiny toes to make tiny thumbs. He rooted up all the tendons, nerves, arteries, bones, so attempted to make a thumb with good sensation and some movements. The eye findings in male patients of Ectrodactyly are very rare.

The association of Ectrodactyly with absence of meibomian glands was noted by Almedia SF[4]. The ocular surface disorders and shortened tear film breakup time in Ectrodactyly were attributed to the absence of meibomian glands, leading to lipid layer deficiency in the tear film with a concomitant increase in tear evaporation[5]. The absence of lacrimal puncta in both eyes of 27 years old woman of Ectrodactyly was seen, when she presented with epiphora and ocular pain[6]. In our study the ocular associations of myopia and cataract were persistently noted in all affected patients of ectrodactyly which have never been reported earlier.

The bilateral cataract is often inherited; autosomal dominant being the most common inheritance pattern[7]. Systemic diseases can cause bilateral cataract and approximately 5% to 10% of bilateral cataract is associated with a systemic disorder[8] e.g. idiopathic (60%), hereditary (30%) autosomal dominant, autosomal recessive or X-linked systemic disorders (5%) Lowe's syndrome, galactosemia, Trisomy, Alport's syndrome, Myotonic dystrophy, Marfan's syndrome etc, also cause bilateral lens pathology. Marfan's syndrome, Homocystinuria and weil-marchesani's syndrome have systemic features and lens anomalies. Marfan's syndrome inherited in an autosomal dominant manner, is characterized by wide spread skeletal abnormalities including Arachno-dactly[9]. The Marfan's syndrome has cardiovascular abnormalities. But none of our study cases had such anomalies[10].

Table 1: Sex wise distribution

Sex	No of patients	%
Male	9	60
Famale	6	40
Total	15	100

Table 2: Eye pathologies (Myopia + Lens changes)

	No of patients	%
Affected	3	20
Unaffected	12	80
Total	15	100

Table 3: Sex distribution of total cases

Sex	No of patients	%
Male	25	54.3
Famale	21	45.7
Total	46	100

Table 4: Affected sex distribution 22 out of total 46 (47.82%)

Sex	No of patients	%
Male	10	21.7
Famale	12	26.1
Total	22	47.8

Homocystinuria inherited in an autosomal recessive manner, is an inborn error of metabolism involving a deficiency of Cystathionine synthetase. These patients typically suffer from vascular thrombotic problems and most of these patients also have ectopia lentis[11]. None of the patients in our study showed such embolic problems. The weil-marchesani's syndrome[12] exhibits a picture of brachycephaly, short stature, ectopia lentis and micro spherophakia, but no such pathology was seen in our study patients.

CONCLUSION

The Ectrodactyly (Lobster Claw Syndrome) is a rare congenital deformity of the hands and feet. Its inheritance pattern is autosomal dominant. It affects both males and females. Typically person with Ectrodactyly has a cleft where the middle finger or toe adopts a condition that gives the hand or foot the appearance of a lobster claw, but Ectrodactyly patients can have any number of unusual arrangements of the digits. But all of our study patients had similar pattern of hands and feet abnormalities and also the same eye findings (myopia + cataract). The association of many ocular problems e.g., punctal atresia, trichiasis, punctate epithelial erosions and nasolacrimal duct atresia, has been described in the literature but no such findings were seen in our patients. The deformity of hands and feet can be treated surgically to improve functions and appearance. The prosthetics may be used and genetic counseling should be given to the parents about the condition. This inheritable deformity does not affect the mental capabilities of the patients rather the affected members are mentally sharp, intelligent and active.

Although the Ectrodactyly is rare autosomal dominant inheritable disease, but may be very rarely sporadic (with no family history of this malformation). The association of the Ectrodactyly with myopia and cataract is also rare. A lot of investigations, genetic workout and screening are required. The treatment of myopia with glasses and cataract with cataract extraction with posterior chamber lens implantation were promising and satisfactory.

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