CASE REPORT

Focal Dermal Hypoplasia (Goltz Syndrome)

Luay Abdulla Al-Nouri

ABSTRACT:

BACKGROUND:

Focal Dermal Hypoplasia (FDH) is a condition of multiple features . It is important to recognize on clinical grounds , as it may show life threatening complications that need to be dealt with by properly timed interventions

CASE REPORT:

A seven year old girl presented with a skin lesion that was present since birth It was distributed on the forehead ,chest, upper abdomen, and buttocks .All four limbs were also involved .It consisted of linear areas of thinning of the skin ,in which there was herniation of fatty tissue in the form of yellow papules, together with dyspigmentation and telangiectasia.

The nails were dystrophic. There was complete syndactyly of the left second and third toes, and partial fusion of the right second and third toes.

The teeth were all defective and many were carious.

DISCUSSION:

There are between 200-300 cases reported in literature , so it is not a rare condition. They are mostly females , as affected males do not usually survive. Papillomas in different sites may be symptomatic and require surgical intervention. It is a sex linked dominant condition associated with mutation of PORCN gene mapped to locus Xp11.23 .

INTRODUCTION:

Focal Dermal Hypoplasia (FDH)is an uncommon ,but not a rare condition as 200 – 300 cases been reported in literature .It is important to recognise and properly follow up since patients may develop life threatening complications which may be avoided by timely surgical intervention.

CASE REPORT :

Ahlam was a seven year old girl from Baghdad who presented with a skin lesion that was present since birth .The mother had a normal pregnancy, apart from a diarrhoeal episode which was treated with sulfaguanidine tablets. She did not have a history of miscarriage . She was born at full term and delivery was normal.

Ahlam looked small at birth,(birth weight unknown), but had no perinatal anoxia or jaundice. She sat unaided at the age of 7 months, walked at the age of 14 months, and had no delay in acquiring speech. She was doing well at school. She was breastfed for three years , but had only one oral polio vaccine and one diphtheria ,tetanus ,pertussis vaccine given.

The father was 65 years old and the mother was 50 years old. They were cousins. She had 4 brothers and 4 sisters all well .All the sisters were married and had children and one of the brothers was married and had children ,but none had any of Ahlams abnormalties.

On examination she was a pleasant cheerful girl , looking small for her age and was actually shorter than her younger brothers (Fig.1)Her height was 103 cm,her weight 12kg and her head circumference 47.5 cm, all were less than 5th centiles. No cardiovascular or respiratory abnormality detected. Blood pressure was 100 /60.No abdominal organomegaly or neurological abnormality found.

The face showed atrophic scars on the left side of the forehead. There was heterochromia iridis with the right iris darker than left and the sclerae mildly blue .Visual acuity was 6 /9 in both eyes .The bridge of the nose was narrow and depressed while the tip was wide and asymmetric .The chin was pointed.(Fig.2)

The skin lesion was dry and thin with reticular pigmentation and dyspigmentation ,and patchy atrophy especially of the depigmened areas .It was distributed on the lower chest ,upper abdomen ,buttocks and the back was relatively spared. On the extremities, the lesion was linear with symmetrical distribution , more on the ulnar aspect of forearms, with lipoid xanthomatous papules due to herniation of fatty tissue through the underdeveloped dermis, together with pigmentations and hypopigmentations(Fig.3) . Small papillomata were found in the perianal region.

Department of Pediatrics, University of Baghdad

In the right hand, the index finger was short due to the middle phalanx being underdeveloped. (Fig4).In the left hand
, the second,third and fourth fingers were of the same length due to hypoplasia of the proximal phalanx of the third finger .All the nails were dystrophic (Fig.4)
There was complete syndactyly of the second and third toes of the left foot,and partial fusion of the second and third toes of the right foot. All the toe nails were dystrophic.(Fig.5)
The teeth were all showing a variable degree of hypoplasia and most of them were carious .One lower incisor was lost in a fall accident ,one other upper incisor was congenitally absent .Radiololgically, some roots of decidous teeth were short ,the crowns of perminant teeth were hypoplastic with delayed eruption together with generalized alveolar bone resorption (Fig.6)
The ears presented no abnormality, and audiograms were normal. Wrist radiograms revealed a bone age of 3 years. Chest X-Rays were normal.
The histopathology of the skin lesion showed a focally thin epidermis with a basal layer irregularly pigmented with increase of melanin in some parts and decrease in others. There was no pigment incontinence. The dermis was fibrosed and thin ,and not showing pigment accumulation .The skin appendiges were poorly developed .The hair follicles were small with abortive hair formation .No well developed sweat glands or sebaceous glands found.

DISCUSSION:

In 1934, Lieberman from Moscow Polyclinic reported a 14 year old girl with hypoplasia of teeth ,areas of linear atrophy , telangiectasia and pigmentation .Histologically the lesion showed partial to complete replacement of the dermis by adipose tissue[(1)].
In 1941, Cole et al reported a woman with hypoplasia of teeth , syndactyly with areas of skin atrophy , pigmentation and telangiectasia . Histologically, showed areas of complete absence of corium[(2)].Goltz et al in 1962 recognized the condition as a separate entity and gave the name of Focal Dermal Hypoplasia[(3)]. Ever since that time 200-300 cases were reported from all over the world, affecting all races, so it is an uncommon but not a rare disorder[(4)].
The condition is a distinct skin anomaly with a variety of defects of eyes ,teeth and skeletal, urinary ,gastrointestinal, cardiovascular and nervous systems anomalies.It is usually but not always ,an X-linked dominant (lethal in males).The genetic defect has been identified as a mutation of the PORCN gene on the X chromosome mapped to locus p11.23.Ninety per cent of patients are females and 10% are males ,accounted for by postzygotic somatic mosaiacism, which is also

postulated for sporadic female cases with negative family pedigree analysis [(4,5,6)].
The clinical features that are described in literature are :
Cutaneous features : The skin lesion is characteristic giving the condition its name ,usually present at birth but may progress and evolve over time.
It is a symmetrical, linear reticulated, frequently tender , pink or red thin skin. The involved areas may be atrophic, slightly raised or depressed macules. The lesions may follow the lines of Blaschko , and can be few millimetres to few centimeters in width .In the pigmented skin, the lesion may be hypo- or hyperpigmented instead of erythematous. There may be ulcerations and telangiectasia. The distribution may be anywhere on the body but more commonly on the thighs , forearms and cheecks.
The dermis may be totally or partially replaced by accumulation of adipose tissue which gives rise to a hernia like outpouching of fatty tissue, a feature which is unique to FDH.
One of the striking features is the appearance of rasberrylike papillomas, which are multiple , often arisng at the junction of mucosa and skin (perioral ,periocular, perianal and perivulvar). Inside the body, they may appear in the the larynx, oesophagus, and stomach leading to obstruction. They less often appear on the ears, fingers, toes ,umbilicus and groin. New papillomas may continue to appear throughout childhood or even adulthood [(4)].
The skin adnexiae may be involved so the hair may be sparse or brittle and may be absent on scalp or pubic area .The nails may show atrophy ,dystrophy ,spooning or grooving. There may be hypohidrosis or anhidrosis,and apocrine nevi.
Facial abnormalities: There may be asymmetry of the face with mild hemiatrophy ,a narrow nasal bridge, broad tip and notched alae nasi on one side. The chin may be pointed[(4)].
The ears may be low set, protruding , asymmetric or malformed with hypoplasia of the halix. Deafness may be conductive or neurosensory. There may cocklear dysplasia. Papillomata may appear at or near external auditory meatus [(4)].
The eyes may be widely spaced with sparse eyebrows. There may be microphalmia ,anophthalmia, strabismus ,or nystagmus. The eyelids may show ectropion ,ptosis ,sparse eyelashes or blocked lacrimal ducts .The sclerae

may be blue and cornea may be defective. There may be heterochromia ,aniridia ,irregular pupils or coloboma of the iris. There may be ectopia lentis ,cloudy vitreous ,coloboma of choroids ,coloboma of retina and neovasculrisation the optic nerve may be hypoplastic.There may papillomas at lid margin or conjunctiva [4,7].

Mouth: There may be prognathism or cleft palate. The palate may be high arched with cleft or papillomas. The gums may be hypertrophied or carry a papilloma. Papillomas may appear on the tongue .The teeth may shaw agenesis, dysplasia, enamel defect or notching. There may be delayed eruption, overbite or malocclusion [4,8].

Skeleton : The patients are of short stature with trunk and limb asymmetry. There may be aplasia of parts of limbs or even amelia. Syndactyly was found in 60% of the cases .There may be adactyly ,clinodactyly ,polydactyly ,split hand or feet or clawhands.

There may be abnormal clavicles or ribs, kyphoscoliosis or pelvic bones hypoplasia.

Bone radiograms may reveal ostiopathia striata in 20%of cases, which is characterstic but not pathognomonic .These are longtudinal striations in the diaphysis ,bilateral and symmetrical in the long and sacral bones. They are vertical opacities originating at articular surfices extending into diaphysis where they narrow and disappear[9,10].

The other systems involved are the cardiovascular in the form of anomalous venous return. Neurologic anomalies include microcephaly , cerebellar atrophy and meningomyelocoele .There may be intestinal malrotation, oesophageal papilloms ,gastric polyps,diaphragmatic hernia ,omphalocele, or rectal prolapse. The kidneys may be hypoplastic, absent or horseshoe kidney .Recurrent respiratory infections may be noticed ,cellulitis ,conjunctivitis, otitis media and urinary tract infection may be present , but no immune system disorder identified[4].

Treatment is usually directed towards soft tissues , teeth, and skeletal anomalies for optimal functional and aesthetic results .Laser treatment may ameliorate pruritus ,erythema or telangiectasia. Papillomas may be removed surgically for aesthetic reason or obstruction of respiratory or gastrointestinal tracts[11].

REFERENCES:

1. Liebermann. S. Atrophoderma linearis maculosa et Papillomatosis congenitalis, Acta Dermato-vener,1935;16:476-84.
2. Cole. H.N., Driver . J.R., Griffen . H.K. , Norris . C.B. , and Strand. G. Ectodermal and Mesodermal Dysplasia with Osseous Involvement .Arch.Derm.Syph.,1941;44:773-88.
3. Goltz.R.W.,Peterson W.C.,Gorlin. R .J.,and Ravits. H.G. Focal Dermal Hypoplasia . Arch.Derm.,1962;86:708-17.
4. Goltz . R.W., Krafchik, B.R., Butler . D.F, Chan,E.F., Crawford . G.H., and Elston D .M. Focal Dermal Hypoplasia . Acta.Derm.Veneriol.,1998;78:235-6(updated July 30,2008)
5. Grazeschik . K .H ., Bornholdt .D , Offner F .,Konig .A , del Carmen Boente .M,Enders, H,et al . Defeciency of PORNC ,a regulatory Wnt signalling is associated with focal dermal hypoplasia. Nat Genet., 2007;39:633-5.
6. Wang . X, ReidSutton.V, Omar Peruza - Llanes, J. Yu .Z ., Rosetta. R , Kou, Y .C .et al .Mutations in X-linked PORSN , a putative regulator of Wnt signalling cause focal dermal hypoplasia.Nat.Genet.2007;39:836-8.
7. Thomas . J.V., Yoshizumi. M.O ., Beyer .C.K. , Craft. .J.L. Alber D .M. . Ocular manifestations of focal dermal hypoplasia,Arch.Ophthalmol.,1977:95.
8. Al-Ghamdi .K, Crawfod .P. J. Focal Dermal Hypoplasia , Oral and dental findings ,J. Clin .Pediatr .Dent. 2005;30:67-72.
9. Knockaert . D , Dequeker . J . Ostiopathia striata and focal dermal hypoplasia ,skeletal radiol. 1979; 4:223-7.
10. Mianda . S .B. , Delmaestro . D , Bertoli . R, Marinhp . T . Lucas . E . Focal Dermal Hypoplasia with exuberant fat herneation and skeletal deformities . Pediatr .Dermatol. 2005;22:420-3.
11. Alster . T.S .,Wilson . F . Focal Dermal Hypoplasia (Goltz syndrome), Treatment of cutaneous lesions with the 585.nm flashlamp-pumped pulsed dye laser . Arch. Dermatol. 1995;131:143-4.