

A Newborn with Goltz Syndrome: Case Report

Goltz Sendromlu Bir Yenidoğan

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ABSTRACT Goltz syndrome (focal dermal hypoplasia) (OMIM #305600) is an X-linked dominant disorder characterized by developmental anomalies of ectodermal and mesodermal tissues. Recently, it was found that Goltz syndrome is caused by mutations in the *PORCN* gene located Xp11.23. Ninety-five percent of the cases have been reported to be sporadic. Focal dermal hypoplasia is a rare disorder characterized by cutaneous lesions including telangiectasias, hypopigmentation, hyperpigmentation, and/or dermal atrophy following Blaschko's lines. In this article, a newborn girl with Goltz syndrome was presented.

Key Words: Focal dermal hypoplasia; infant, newborn

ÖZET Goltz sendromu (fokal dermal hipoplazi) (OMIM #305600), ektodermal ve mezodermal dokuların gelişimsel anomalilerin görüldüğü X bağlı dominant geçiş gösteren bir hastalıktır. Son zamanlarda, Goltz sendromuna Xp11.23'te lokalize *PORCN* genindeki mutasyonların yol açtığı bulunmuştur. Olguların %95'i sporadik olarak bildirilmiştir. Fokal dermal hipoplazi Blaschko çizgilerini izleyen telanjiektazi, hipopigmentasyon, hiperpigmentasyon ve/veya dermal atrofiyi içeren kutanöz bulgularla karakterize nadir bir hastalıktır. Bu makalede, Goltz sendromlu yenidoğan bir kız bebek sunulmuştur.

Anahtar Kelimeler: Fokal dermal hipoplazi; bebek, yenidoğan

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Goltz syndrome (focal dermal hypoplasia, FDH) (OMIM #305600) is an X-linked dominant disorder characterized by developmental anomalies of ectodermal and mesodermal tissues.¹ Recently, it was found that FDH is caused by mutations in the *PORCN* gene located Xp11.23.² Ninety-five percent of the cases have been reported to be sporadic.³ FDH is a rare disorder characterized by cutaneous lesions including telangiectasias, hypopigmentation, hyperpigmentation, and/or dermal atrophy following Blaschko's lines.⁴ Associated variable features include patchy or linear areas of hairlessness, periorificial papillomas, hypoplasia or aplasia of bones resulting in asymmetric appearance of the face and the body, malformations such as syndactyly or aplasia of bones, coloboma, and microphthalmia or unilateral anophthalmia. In addition, hypodontia or

oligodontia, hypoplasia of the enamel, hearing loss, myelomeningocele, bifid ureter, horseshoe kidney, omphalocele or papillomatosis of the larynx may be found.⁵ In this article, a newborn girl with Goltz syndrome was presented.

CASE REPORT

A 1-day-old girl was admitted to the newborn intensive care unit because of skin lesions and multiple skeletal anomalies. The pregnancy was uneventful. She was at term, live born infant. She was the 3rd child of a nonconsanguineous couple. Her physical examination revealed: weight: 2.230 grams (<10th percentile); height 45 cm (<10th percentile); head circumference: 30 cm (<10th percentile); pulse rate: 128/min; respiratory rate: 42/min; blood pressure: 65/45 mm/Hg and temperature: 37 °C. She had facial asymmetry, preauricular skin tag, low-set ears and cleft tongue. She also had an umbilical hernia. She had split hand malformation. The right first and second toes were fused and the third toe was absent. Cutaneous examination showed multiple atrophic erythematous lesions of varying sizes on the anterior right chest and extremities along the lines of Blaschko, resembling striae (Figure 1 A, B, C). Ophthalmologic examination did not reveal any abnormalities.

Laboratory examinations including complete blood count, serum electrolytes, liver function tests were all within normal limits. The patient's thyroid function was within normal limits. Karyotype was normal, 46,XX. Echocardiography revealed a small patent ductus arteriosus with a left-right shunt and secundum atrial septal defect. Ultrasonography of the abdomen showed that the right kidney was located in the pelvic fossa, but the left kidney was visible in the left renal fossa and renal ultrasonography revealed a mild enlargement (8 mm) in the left renal pelvicalyceal system due to ureteropelvic junction obstruction. Radiographies of long bones did not reveal striated osteopathy. Radiographies of chest, hands and feet showed the right clavicular hypoplasia and osseous syndactylies. An otoacoustic emission test revealed the hearing loss of the right ear. Magnetic resonance imaging of the brain was normal.

Informed consent was obtained from the parents. Skin biopsy taken from affected skin on the right anterior chest showed a marked dermal hypoplasia with adipose tissue high in the papillary dermis consistent with FDH. The diagnosis of FDH in association with pelvic kidney, atrial septal defect and clavicular hypoplasia was made on the basis of the patient's clinical features and histopathological findings (Figure 2).

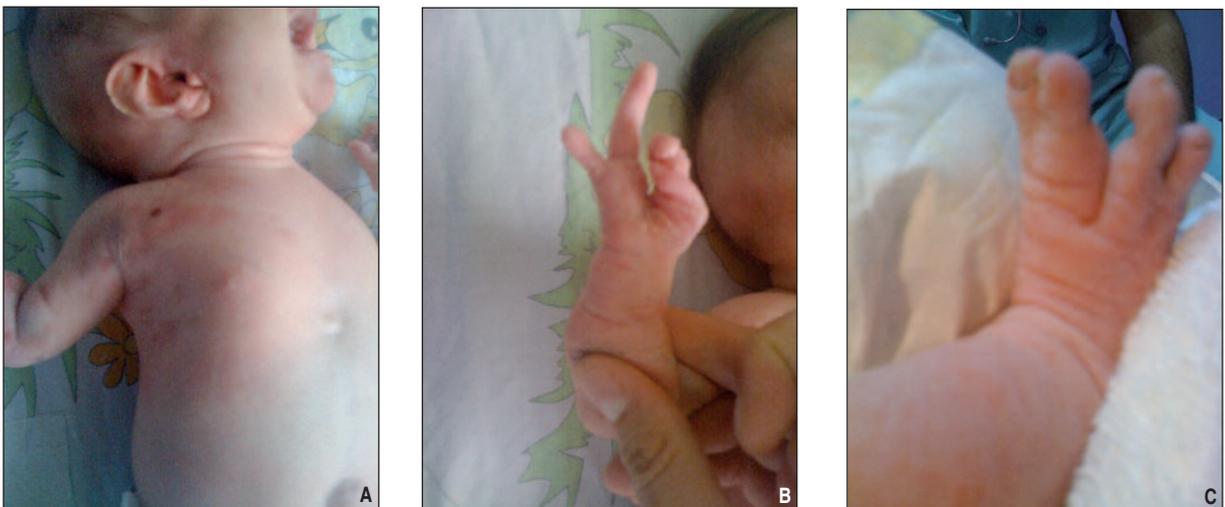


FIGURE 1 A, B, C: 1-day-old newborn with Goltz syndrome: multiple skin lesions on anterior right chest and extremities. She had split hand malformation. The right first and second toes were fused and the third toe was absent.

(See for colored form <http://pediatri.turkiyeklinikleri.com/>)

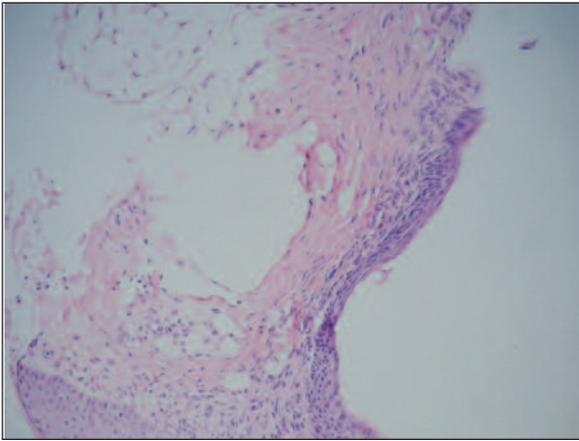


FIGURE 2: Haematoxylin and eosin, (X40). Marked dermal hypoplasia with adipose tissue high in the papillary dermis.

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DISCUSSION

FDH was first recognized as a separate clinical entity by Goltz et al.⁶ In FDH, there is a broad spectrum of developmental ectoderm and mesoderm anomalies which has been reviewed extensively by Goltz et al. and Warburg.^{7,8}

There are a few cases with FDH who were diagnosed in the neonatal period in the literature. We report a newborn with clinical features of FDH.

The most common striking features in FDH are the skin lesions. These include atrophy and linear pigmentation of the skin, herniation of fat through the attenuated dermis, multiple papillomas of the mucous membranes or skin, especially that of the lips, gums, arms, and vulva, sparse scalp hair with small areas of localized alopecia, and dystrophic nails.⁴ Our patient had multiple atrophic erythematous lesions of varying sizes on the anterior right chest and extremities along the lines of Blaschko.

Skeletal defects are the second most common abnormality in FDH. Skeletal anomalies include asymmetric involvement of the hands and feet in 60% of patients, including syndactyly, ectrodactyly, polydactyly, absence or hypoplasia of digits, and even absence of an extremity. Cervical rib has been reported.⁹ Skeletal asymmetry, clavicular dysplasia and spina bifida occulta can occur.⁷ Our patient had multiple skeletal anomalies and clavicular hypoplasia.

The eye findings most frequently noted are colobomas, microphthalmia and strabismus. The teeth are commonly affected by enamel defects with caries, dysplasia, irregular spacing and malocclusion. These individuals tend to be in the low normal range for height and weight.¹⁰ In our patient, ophthalmologic examination was normal. However, she was small for gestational age.

Occasional anomalies include joint hypermobility, mental retardation, hearing defects, microcephaly, horse-shoe kidneys, umbilical, epigastric or diaphragmatic hernias.¹¹ Of all these features, our patient had microcephaly, umbilical hernia and hearing defect.

Han et al. described an infant girl of 36 weeks' gestational age who had cardiovascular and other lethal internal anomalies in addition to characteristic external anomalies of FDH.¹² The internal anomalies included truncus arteriosus type II with truncal origin of hypoplastic pulmonary arteries, cardiac ventricular septal defect, severe hypoplasia of the lungs and pulmonary veins, massive diaphragmatic hernia, and absence of the right kidney. Irvine et al. reported a child with typical cutaneous, ocular, and skeletal manifestations of FDH, who had also preauricular sinuses, omphalocele, duodenal atresia, mediastinal dextraposition, patent ductus arteriosus, esophageal reflux, and hydronephrosis.¹³ However, our patient had a pelvic kidney, congenital heart disease including secundum atrial septal defect.

The main condition from which FDH must be differentiated in early life is incontinentia pigmenti although Rothmund-Thomson syndrome must also be considered.¹⁴

FDH sometimes initially shows an inflammatory phase in which there may also be blistering and crusting. Macular areas of hypopigmentation and hyperpigmentation also occur. Skin lesions tend to be linear. Forty per cent of FDH patients have ocular abnormalities and 80% have skeletal abnormalities.¹⁴ These physical signs all occur in incontinentia pigmenti where they are frequently accompanied by blood leucocytosis with eosinophilia. The diagnosis of FDH can be made

when the hypoplastic affected areas of skin with characteristic histology are found.¹⁵

Neonatal Rothmund-Thomson syndrome shows linear indurated erythematous lesions with photosensitivity. These lesions subside leaving shiny atrophic skin with telangiectasia. Macular hyperpigmentation may be present as may other congenital ectodermal and skeletal defects. The distinction from FDH must be made on histologic grounds.¹⁵

In Rothmund-Thomson syndrome the epidermis is abnormal and shows atrophy, dyskeratosis and basal layer abnormalities, none of which occur in the FDH.⁷

In our patient, the diagnosis was initially suggested by the skin lesion and malformations of the distal extremities. Examination of the skin revealed linear atrophic lesions distributed along Blaschko lines. These observations prompted us to consider FDH as a possible diagnosis, which was subsequently confirmed by histopathological investigation.

In conclusion, FDH is a mesoectodermal disorder manifested by noticeable skin changes at birth with progression thereafter. This diagnosis should be considered in any newborn presenting with multiple atrophic erythematous lesions of varying sizes on skin along the lines of Blaschko.

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