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CASE REPORT

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Rijedak slučaj oralne juvenilne hijaline fibromatoze

Oral Juvenile Hyaline Fibromatosis: A Rare Entity

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Sažetak
Juvenilna hijalina fibomatoza iznimno je rijedak poremećaj kod dojenčadi i djece, a javlja se prema zakonima autosomno recesivnog nasljeđivanja. Izgleda poput multiple kožne ili subkutane tumorne tvorbe, sporo se razvija i češća je u području glave i vrata te gornjeg dijela trupa. Često je povezana s gingivnom hipertrofijom, teškom fleksularnom kontrakturom udova i koštanom lezijom. Nema mentalne retardacije. Histološki se te lezije sastoje od obilne eozinofilne osnovne tvari s neravnomjerno raspršenim fibroblastima. Ekscidirane lezije u ranim stadijima bogatije su stanicama. Točno podrijetlo eozine hijaline tvari nije poznato. Nedavno je pronađen defekt kromosoma 4q21 povezan s lokusom gena – 2 za kapilarnu morfogenezu. Diferencijalna dijagnoza za juvenilnu hijalinu fibromatozu uključuje i infantilnu sistemsku hijalinozu, za koju se zna da je alelna. Trenutačno nema široko prihvaćene učinkovite terapije ni za juvenilnu hijalinu fibromatozu ni za infantilnu sistemsku hijalinozu. Juvenilna hijalina fibromatoza i infantilna sistemska hijalinoza ponekad se teško razlikuju jer su vrlo slične. Mi izvještavamo o slučaju juvenilne hijaline fibromatoze kod 10-godišnje djevojčice s pretežno gingivalnom hiperplazijom.

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Uvod

Juvenilna hijalina fibromatoza (JHF) rijetka je autosomno recesivna bolest kod koje se stvaraju i odlažu hijaline tvari u koži i drugim organima (1,2).

U literaturi su opisana dva oblika: juvenilna hijalina fibromatoza i infantilna sistemska hijalinoza (ISH). Oba se sindroma nasljeđuju autosomno recesivno te imaju slične kliničke i histopatološke nalaze (1, 2). JHF se javlja tijekom nekoliko prvih godina života i većina oboljelih preživi do odrasle dobi – najstariji preživjeli pacijent u dobi je od 51 godine (1). ISH se javlja u prvim tjednima života te pacijent obično živi samo do 20 mjeseci (3**).** Klinička slika ISH teža je i sastoji se od razbacanih kožnih manifestacija, ponovljenih infekcija i dijaree. Djeca uglavnom umiru, a ona s JHF-om imaju dulji životni vijek s manje ozbiljnim manifestacijama (4). Dosad su se JHF I ISH opisivali kao dva odvojena entiteta, a danas se smatraju dijelom spektra iste bolesti jer je nedavno u genetičkim istraživanjima pronađena kod pacijenata mutacija istog gena (1). Hutchinson je opisao slučaj JHF-a povezanog s teškom gingivnom hipertrofijom te istaknuo kako u literaturi ima premalo opisa takvih slučajeva (3). Mi izvještavamo o slučaju JHF-a koji se očitovao uglavnom kao lokalizirano povećanje desni.

Introduction

Juvenile hyaline fibromatosis (JHF) is a rare autosomal recessive disease characterized by the production and deposition of hyaline material in the skin and other organs (1, 2). Two distinct forms have been described in the literature: juvenile hyaline fibromatosis and infantile systemic hyalinosis (ISH). Both syndromes are autosomal recessive conditions presenting with similar clinical and histopathological finding (1, 2). JHF occurs within the first few years of life, and most patients described have survived into adulthood, with the oldest reported survivor being 51 years old, (1) whereas ISH occurs in first few weeks and patient generally survives only up to 20 months (3). The clinical presentation of ISH is more severe and presents with diffused skin manifestations, involvement of joint infections, recurrent infections, and diarrhea. It has a fatal outcome whereas JHF has prolonged life span with less severe manifestations (4). Previously JHF and ISH were described as two distinct entities, but they are now considered a part of the same disease spectrum, as recent genetic studies have demonstrated mutations in the same gene in these patients (1). Hutchinson reported a case of JHF associated with severe gingival hypertrophy and mentioned lack of dental literature for this entity (3). We report a case of JHF which presented mainly as localized gingival enlargement.

Opis slučaja

U ordinaciju oralne medicine i radiologije došla je 10-godišnja pacijentica s pritužbom da joj se u posljednja dva mjeseca progresivno povećava oteklina u prednjem mandibularnom području te kako ne može žvakati na oboljeloj strani. Intraoralnim pregledom bila je otkrivena gingiva koja se protezala od desnog inciziva do desnog očnjaka i desnih mliječnih kutnjaka, uključujući zube 41,42,43,44. Izraslo tkivo prekrivalo je krune i bilo je po konzistenciji tvrdo te veliko 1,5 x 2cm (Slika 1.).

Lezije su na površini imale otiske zuba antagonista. Submandibularni limfni čvorovi bili su povećani i osjetljivi na palpaciju. Pacijentica je imala također višestruke asimptomatske i hiperpigmentirane izrasline na licu i u području uha veličine oko 0,5 milimetara, čvrste konzistencije te vezane za fasciju (Slika 2). Obiteljska anamneza otkrila je blisko krvno srodstvo roditelja. Trudnoća majke bila je uredna i završila je redovitim porođajem bez komplikacija. Pacijentičina 8-godišnja sestra nije bila pogođena. Temeljito ispitivanje roditelja i članova obitelji nije otkrilo slične slučajeve u bližoj ili široj obitelji. Temeljiti pregled nije otkrio druge abnormalnost na ostalim dijelovima tijela. Radiološki nalaz mandibule pokazao je ultilokularnu radiolucenciju u području desno od simfize te proširene bukalne kortikalne ploče (Slika 3.). Rutinski hemalaumeozinski preparat otkrio je blagu anemiju, a drugi biokemijski testovi bili su u sklopu normalnih granica.

Postavljena je privremena dijagnoza idiopatske gingivalne fibromatoze i lezija gigantskih stanica. Bila je obavljena i incizijska biopsija. Roditelji pacijentice odbili su biopsiju lezija na licu i uhu.

Histološki nalaz

Histološki rezovi pokazali su tumornu masu bogatu staničnim elementima i mnogo homogene osnovne tvari koja je sadržavala ravnomjerno raspoređene okrugle, nezrele fibroblaste (Slika 4.). Stanice su bile vakuolizirane te u nekim dijelovima blizu retrakcijskih artefakata. Uočena su mnoga prokrvljena područja tankih stijenki s RBC-om (Slike 5. i 6.). Poneke stanice kao da su bile poredane oko razgranatih krvnih žila. Postavljena je konačna dijagnoza juvenilne hijaline fibromatoze.

Rasprava

JHF je iznimno rijedak nasljedan poremećaj s povećanjem gingive, kožnih čvorova i fleksuralne kontrakture velikih zglobova (5). Prvi ju je opisao Murray 1873. godine i nazvao "mollusum fibrosum" (6, 7). Izrazom JHF prvi se koristio Kitano sa svojim suradnicima 1972. (5). Poznat je prema mnogim termnima, poput juvenilna hijalinoza, fibromatosis hyalinica multiplex, Murray-Paretic-Drescherova sindrom (8).

Lezije se karakteristično nalaze na koži, na gingivi kao hiperplazije, te kod bolesti zglobova i kosti sa sistemskim zahvaćanjem organizma. Koštane lezije deformiraju, ali su asimptomatske. Obično postoje dvije vrste koštanih lezija –

Case report

A 10 year old female patient was referred to out patient department of oral medicine and radiology with chief complaint of progressively increasing swelling in the mandibular anterior region of the jaw for 2 months and inability to eat on the affected side. Intraoral examination showed gingival growth extending from right incisor, canine to right deciduous molar region involving 41,42,43,44. The growth seemed to cover the crown of the teeth and was firm in consistency measuring about 1.5 x 2 cm (Figure 1). The lesion showed indentation of the opposite dentition on the surface. Submandibular lymph nodes were enlarged and tender on palpation. Patient also presented with multiple growths on the face and ear region which were asymptomatic and showed hyperpigmentation measuring about 0.5mm, firm in consistency, bound to the underlying fascia (Figure 2) . Family history revealed consanguineous marriage of parents. The pregnancy of the mother was unremarkable and had a normal delivery. The patient's younger sister aged 8 was unaffected. A thorough questioning of the parents and relatives of the patient failed to give any other family members with similar history or findings. A thorough examination of the patient did not reveal any abnormality in other parts of the body. Radiographic finding of the mandible showed multilocular radiolucency in mandibular right symphyseal region, along with expansion of buccal cortical plate (Figure 3). Routine haematological examination revealed mild anaemia and other biochemical tests were under normal limits.

Provisional diagnosis of idiopathic gingival fibromatosis and giant cell lesion was made. Incisional biopsy of the lesion was carried out. Patient refused biopsy of lesions on face and ear.

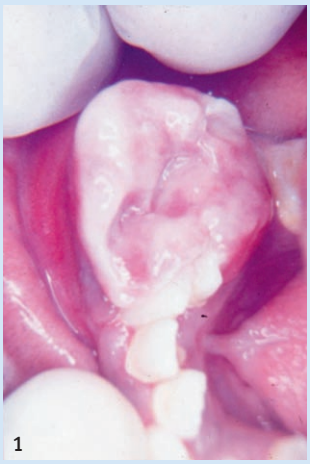
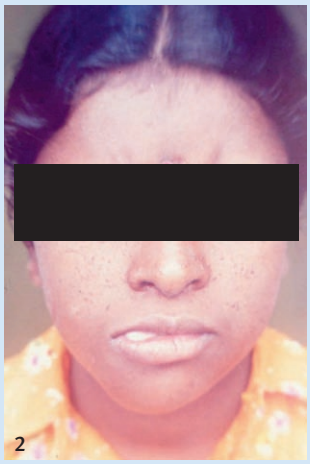
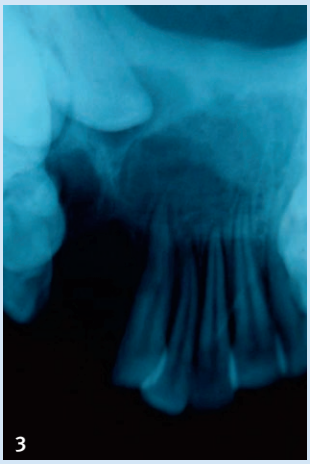
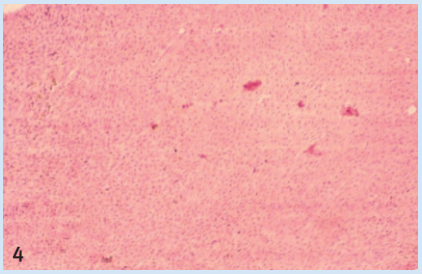
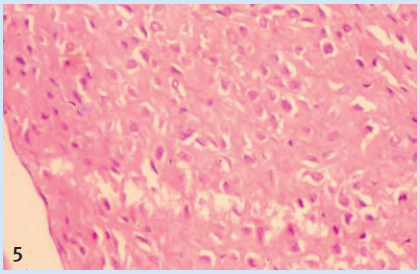
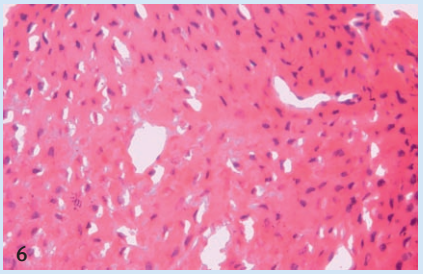
Histological findings

On histological examination, the section showed highly cellular tumor mass composed of copious eosinophilic, homogenous ground substance containing uniformly dispersed plump, round immature fibroblasts (Figure 4). The cells also showed a vacuolated appearance in some areas with adjacent retraction artifacts. Many thin-walled vascular spaces with RBCs were seen (Figure 5, 6). Some cells appeared to be arranged around prominent branching blood vessels. The final diagnosis of "Juvenile hyaline fibromatosis" was made.

Discussion

JHF is an extremely rare hereditary disorder characterized by gingival enlargement, cutaneous nodules and flexural contractures of the large joints (5). It was Murray in 1873 who first described JHF and termed it "mollusum fibrosum" (6, 7). The term JHF was given by Kitano et al. (1972) (5). It is also known as juvenile hyalinosis, fibromatosis hyalinica multiplex, Murray-Paretic-Drescher syndrome (8).

The condition characteristically presents as skin lesions, gingival hyperplasia, joint and bone disease with systemic involvement. The skin lesions are disfiguring but asymptomatic. Two types of skin lesions are usually present: pink pearly papules or plaques which are commonly present on the chin,

Slika 1. Intraoralna slika fibroznog, egzofitičnog tkivnog izrasta na alveoralnom grebenu
Figure 1 Intraoral photograph of a fibrous, exophytic growth on the alveolus.
Slika 2. Ekstraoralna fotografija s prikazom kožnih lezija na licu
Figure 2 Extraoral photograph showing skin lesions on the face.
Slika 3. Radiografija s prikazom dobro ograničene multilokularne radiolucencije
Figure 3 Radiograph showing ill defined multilocular radiolucency.
Slika 4. Mikrofotografija koja prikazuje stanično vezivno tkivo s hijalinizacijom (H i E, 10X).
Figure 4 Photomicrograph showing cellular connective tissue with hyalinization (H&E, 10X).
Slika 5. Mikrofotografija pokazuje stanične elemente u leziji s hijalinizacijom strome (H i E, 40X).
Figure 5 Photomicrograph showing cellularity in the lesion with hyalinization of the stroma (H&E, 40X).
Slika 6. Mikrofotografija s prikazom karakteristične reakcije artefakta oko staničnih jezgri (H i E, 100X).
Figure 6 Photomicrograph depicting characteristic retraction artifact around the nuclei (H&E, 100X).

jedna kao ružičaste biserne papule ili plakovi koji se obično nalaze na bradi, nazolabijalnom naboru, čelu, ušima, stražnjem dijelu vrata i perianalnom području te druga kao veliki subkutani tumori u području vlasišta, a rjeđe na trupu, udovima ili vjeđama (9).

Bolest sporo napreduje i za nju je karakteristično nehemoragično, tvrdo, bezbolno povećanje maksilarne ili mandibularne gingive. Hiperplastično tkivo normalne je boje i nije osobito sklono fizičkoj traumi. Stupanj hiperplazije može varirati od blage do opsežne te se protezati do okluzalne površine ili incizalnog brida zuba. Zbog veće debljine i promijenjenih fizičkih karakteristika desni, denticija može kasniti s erupcijom. Povećanje obično počinje nicanjem trajne denticije, povremeno nicanjem mliječne denticije, a rijetko je kod rođenja. Stanje se može činiti generaliziranim procesom koji uključuje oba zubna luka – mandibularni i maksilarni – ili može biti nepotpun te uključivati samo lokalizirana područja pojedinih zubnih lukova ili se razvijati u dobi od 10 do 35 godina.

Najčešća lokalna prezentacija gingivalne fibromatoze smještena je na palatalnoj strani maksilarnog tubera i lingvalnoj strani mandibularnog alveolarnog grebena. Takva povećanja desni mogu biti jednostrana ili obostrana te se tada na-

nasolabial folds, forehead, ears, back of the neck and perianal region and large subcutaneous tumors found mainly on the scalp and less frequently on the trunk, extremities and eyelids (9).

The disease is characterized by a slowly progressive, nonhemorrhagic, firm, painless enlargement of the maxillary **or** mandibular gingiva. The hyperplastic tissue is of normal color and is not especially prone to physical trauma. The degree of hyperplasia may vary from mild to severe, extending on to the occlusal and incisal surfaces of the teeth. Because of the increased thickness and physical nature of the gingiva, the dentition may be delayed in eruption. The enlargement usually begins with eruption of the permanent dentition – occasionally with eruption of the deciduous dentition – and is rarely present at birth. The condition may appear as a generalized process involving both maxillary and mandibular arches or it may be incomplete, involving only localized areas of either arch or characteristically develops between the ages of 10 and 35 years. The most common local presentation of gingival fibromatosis is on the palatal aspect of the maxillary tuberosities and the lingual aspect of mandibular alveolus. These localized gingival enlargements may be unilateral or bilateral and are called symmetrical fibromas. How-

zivaju simetrični fibromi, no moramomistaknuti nisu pravi tumori. Witkop je predložio izraz simetrična gingivna fibromatoza (10).

Naš slučaj s lokaliziranom gingivnom hiperplazijom sliči slučaju H. Usla koji je opisao slučaj 9-godišnje djevojčice s blagom gingivnom hiperplazijom (11). James J. Scuibba predstavio je slučaj JHF-a s generaliziranim povećanjem gingive te samo dva podkožna čvorića na čelu (10). Naš slučaj također karakterizira lokalizirana oteklina desni na alveolarnom nastavku mandibule te mali hiperpigmentirani čvorići na licu i uhu, bez lezija mekih tkiva ili na ostalim dijelovima tijela.

Histopatološki su lezije sadržavale homogenu amorfnu eozinofilnu osnovnu tvar, koja je donekle nalikovala na hrskavično tkivo. Vretenasti fibroblasti bili su poredani u trakastom rasporedu. Udjel stanica u odnosu prema osnovnoj tvari varira od jednog do drugog tumora (5). Podrijetlo i priroda hijaline tvari još nisu točno poznati (1). Prema dosadašnjim spoznajama uzrok je možda fokalna sinteza glikozaminoglikana i glikoproteina u fibroblastima (1,2,10) s povećanim količinama hondroitin-sulfata i smanjenom razinom hijaluronske kiseline (10). Nedavo je na oboljeloj koži bio pronađen lanac pro-alpha$_2$ i kolagen tipa III, što sugerira poremećaj metabolizma kolagena s povećanjem sinteze i degradacije kolagena tipa I sa sistemskim snižavanjem metabolizma kolagena tipa III. Nekoliko autora smatra da perivaskularno odlaganje hijaline tvari upozorava na intravaskularno podrijeklo, a mehanički čimbenici su „okidači" procesa te ga prati poremećaj regulacije mehanizama popravljanja nakon mikrotrauma (4).

Diferencijalna dijagnoza uključuje ISH, neurofibromatozu, idiopatsku gingivnu hiperplaziju, nodularnu amiloidozu, kongenitalnu generaliziranu fibromatozu, Winchesterov sindrom i lipoidnu proteinozu te mukopolisaharidozu. Idiopatska gingivalna hiperplazija nije povezana s kožnim lezijama. Winchesterov sindrom uvijek je povezan sa zamućenjem kornee. Lipoidna proteinoza je odlaganje amorfne eozinofilne, hijaline tvari u dermisu kože s lukovičastim rasporedom oko krvnih žila. Mukopolisaharoidoza može se razlikovati na temelju histoloških i biokemijskih pokazatelja (8). ISH, kako klinički tako i histološki, nalikuje na JHF. Glavna razlika je da kod ISH lezije pokazuju znakove difuzno zadebljale kože, javljaju se izbočene hiperpigmentirane naslage, uključeni su i unutarnji organi, pacijent ima neprekidan poljev i teške infekcije te na kraju umire. JHF karakteriziraju veliki čvorići obično u vlasištu i pacijenti općenito imaju dulji životni vijek. Klinički JHF i ISH uključuju papularne i nodularne kožne lezije, gingivnu hiperplaziju, kontrakture zglobova i različite stupnjeve ozljeda kostiju (4). Unatoč svemu navedenome u literaturi, stručnjaci se ne slažu u vezi s tim jesu li JHF i ISH dva različita poremećaja ili su dvije varijante iste bolesti (1). Oba poremećaja javljaju se na alelima i opisana su u literaturi kao prijelazni oblici (8). Nalazi svjetlosne i elektroničke mikroskopije upućuju na to da ISH jako nalikuje na JHF, premda kod ISH postoje široke nakupine hijaline tvari, osim kože u organima poput crijeva, nadbubrežne žlijezde, mokraćnog mjehura, skeletalnim mišićima, timusu i paratiroidnim žlijezdama (1). Dosad se terapija lezija JHF-a sastoever, since this lesion is not a true tumor, Witkop proposed the term symmetrical gingival fibromatosis (10).

Our patient presented with localized gingival hyperplasia which is similar to the case reported by Uslu, who reported a 9 year old girl with mild gingival hyperplasia (11). Scuibba reported a case of JHF which mainly presented as generalized gingival enlargement with only two subcutaneous nodules on the forehead (10). Our patient also presented with localized gingival swelling on the mandibular alveolus with a few small hyperpigmented nodules on the face and the ear and failed to show any bony lesions or soft tissue lesions on any other parts of the body.

Histopathologically the lesion shows homogenous amorphous eosinophilic ground substance, sometimes giving a chondroid appearance. Spindle shaped fibroblasts are present in a streak-like pattern. The ratio of cells to ground substance varies from one tumor to another (5). The origin and the nature of hyaline material are still unknown (1). According to recent research, the focal synthesis of glycosaminoglycan and glycoproteins by fibroblasts (1, 2, 10) results in an increase in the amount of chondrotin sulphate and a decrease in hyaluronic acid (10). Recently an absent pro-alpha$_2$ chain and an absent collagen type III chain have been reported in affected skin, suggesting a deregulated metabolism of collagen with increased synthesis and degradation of type I collagen and also with reduced overall metabolism of type III collagen. Some authors even suggest perivascular deposition of hyaline material indicating an intravascular origin of it, mechanical factors being triggers of the process, accompanied by a deregulation of repair mechanism after microtrauma (4).

Differential diagnosis includes ISH, neurofibromatosis, idiopathic gingival hyperplasia, nodular amyloidosis, congenital generalized fibromatosis, Winchester syndrome and lipoid proteinosis, mucopolysaccharidosis. Idiopathic gingival hyperplasia is not associated with cutaneous lesions. Winchester syndrome is always associated with corneal clouding. Lipoid proteinosis demonstrates a deposition of amorphous eosinophilic, hyaline material within the dermis with an onion skin arrangement around blood vessels. Mucopolysaccharidosis can be differentiated on the basis of histological and biochemical findings (8). ISH resembles JHF both clinically and histologically. The main differentiating features between ISH and JHF are: the former shows diffuse, thickened skin, hyperpigmented plaques on bony prominence, visceral involvement, persistent diarrhea, frequent severe infections, and fatal outcome, whereas the latter presents with large nodules which are commonly located on the scalp, the patient generally has a more prolonged life span. The common clinical features of JHF and ISH include papular and nodular skin lesions, gingival hyperplasia, joint contractures and diverse degrees of bone involvement (4). Thus significant controversy exists in the literature as to whether JHF and ISH truly represent two distinct disorders or rather two ends of the same disease spectrum (1). Both disorders are allelic and intermediate phenotypes between these two disorders have been described (8). Light and electron microscopic findings in ISH closely resemble the ones in JHF. However,

jala od kirurškog uklanjanja tumora iz kozmetičkih razloga, premda može biti mutirati i recidivi su česti (1). Za hiperplaziju gingive preporučuje se gingivektomija, no nakon toga obično slijedi povratak bolesti. Uslu i njegovi suradnici pratili su jedan slučaj JHF-a te nakon dvije godine ustanovili da se bolest nije ponovila. Zaključili su da odgovarajući kirurški zahvat osigurava uspješan ishod terapije toga poremećaja, kako estetski tako i funkcionalno (11). Potrebna su daljnja istraživanja kako bi se otkrio način na koji nastaje gingivna hiperplazija i druge abnormalnosti kod JHF-a, tako da se očekivani životni vijek pacijenta može još produljiti.

Mišljenja o kirurškom zbrinjavanju kontraktura zglobova podijeljena su – jedni smatraju da je operativno opuštanje kontraindicirano jer može rezultirati aktivacijom hijaline fibromatoze, a drugi ističu da je indiciran kirurški pristup ako se blagom fizikalnom terapijom ne uspije poboljšati raspon pokreta (1).

U literaturi je samo jedan opisani slučaj JHF-a s komplikacijom oralnog karcinoma skvamoznih stanica nepca i to kod 45-godišnje žene. Unatoč dobrom odgovoru na onkološku terapiju, pacijentica je umrla od aspiracijske upale pluća (5). JHF i JSH ostaju stigmatizirani, oni onesposobljavaju pacijente i ponekad su smrtonosni poremećaji bez odgovarajuće terapije.

Na kraju, JHF ostaje smrtonosna bolest bez prave terapijske mogućnosti. Premda otkriće odgovornog gena otkriva osnovu bolesti, prognoza je loša.

U budućim istraživanjima trebalo bi se usredotočiti na genetsko savjetovanje obitelji te bi kod genetske terapije trebalo ciljati na specifičan gen-uzročnik. U predstavljenom slučaju nije se mogla ustanoviti prognoza pacijentice jer potječe iz nepismene i siromašne obitelji. Unatoč potankom objašnjavanju tijeka bolesti i koliko je važno pratiti bolest, pacijentica nije dolazila na kontrole ni na daljnju terapiju.

there is widespread deposition of hyaline material in the organs outside the skin, in ISH, such as gut, adrenals, urinary bladder, skeletal muscles, thymus and parathyroids (1).

Until now, treatment of the skin lesions in JHF has consisted of surgical removal of the tumors for cosmetic reasons. However, this can be mutilating and recurrences are frequent (1). For gingival hyperplasia, the recommended treatment regime is gingivectomy, which is usually followed by a recurrence of disease. Uslu et al. monitored a gingivectomy in a case of JHF with 2 years follow up; there was no recurrence. They concluded that appropriate surgery insures a successful outcome of treatment for this disorder, both esthetically and functionally (11). Further research is necessary to reveal the underlying mechanisms of gingival hyperplasia and other abnormalities of JHF so that the life expectancy of the patient can be prolonged.

Opinions regarding the surgical management of joint contractures are divided; some suggest that operative release is contraindicated, as it can result in activation of hyaline fibromatosis. In contrast, some believe that when gentle physical therapy fails to improve the range of motion, surgery is indicated (1).

In literature there is only one reported case of JHF complicated with oral squamous cell carcinoma of the palate in a 45 year old female. In spite of a good response to treatment for oral squamous cell carcinoma, the patient died of aspiration pneumonia (5). JHF and ISH remain stigmatizing, incapacitating and sometimes fatal disorders with no satisfactory treatment.

In conclusion, JHF remains a fatal disease with no satisfactory treatment. Even though discovery of responsible gene mutations provides a basis for the disease, the prognosis is poor. Thus, genetic counseling of the families and treatment should be targeted to the gene specific by gene therapy in future research. In the present case, the prognosis of the patient could not be determined as the patient and her parents hail from illiterate and economically backward family. In spite of a thorough explanation regarding the disease process, the patient was lost for follow up and also for further treatment.

Abstract
Juvenile hyaline fibromatosis is an exceedingly rare disorder of infants and children which appears to have autosomal recessive inheritance. It is characterized by multiple, slowly growing dermal or subcutaneous tumors, especially in the head and neck region and upper trunk, often associated with gingival hypertrophy, severe flexural limb contractures and bone lesions. There is no mental retardation. Histologically, these lesions are composed of copious eosinophilic, homogenous ground substance with unevenly dispersed fibroblasts. Lesions excised in early stages are more cellular. The precise nature of the eosinophilic hyaline material is not known. Recently, a defect on chromosome 4q21 associated with the locus of the capillary morphogenesis gene – 2, has been demonstrated. The differential diagnosis of juvenile hyaline fibromatosis includes infantile systemic hyalinosis, which is now known to be allelic. Currently, no widely accepted effective treatment exists for juvenile hyaline fibromatosis or infantile systemic hyalinosis. Infantile systemic hyalinosis and juvenile hyaline fibromatosis are sometimes difficult to separate since they show significant overlap. We report one such unusual case of juvenile hyaline fibromatosis in a 10 year old female presenting mainly with gingival hyperplasia.

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Key words
Fibromatosis, Gingival; Hyalinosis, Systemic; Chromosome 4, trisomy 4q21; Gingival Hyperplasia

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