Review Article

Incontinentia Pigmenti: A Case Report and Literature Review

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Abstract

Incontinentia pigmenti (IP; OMIM#308300) is a rare multisystem disorder with an incidence of 0.7:100,000 live births. IP is rare and predominantly seen in females. Mutations in *IKBKG* gene (Xq28, GenBank: NM_003639.3, OMIM#300248) were reported as underlying cause of IP. *IKBKG* encode NFkB protein, which controls the expression of other genes involved in cell proliferation, immunity, and inflammation. Oro-dental abnormalities have been documented in 50%–75% IP cases. We present a case report of a 16-year-old female with probable clinical IP, Arnold–Chiari malformation, hydrocephalus, delayed psychological development, and seizures. The IP hallmark feature: hyperpigmentation of the skin along the Blaschko's lines was present only on the left side of the body. Left-sided hemifacial and tongue hypertrophy were present, which have not been reported previously. Consistent with published reports, tooth size and shape discrepancies were present. However, unlike previous reports, discrepancies were prominent on affected versus unaffected side. This paper provides IP literature review, clinical considerations, and insight on management.

Keywords: *Blaschko's lines hemifacial hypertrophy, Bloch–Sulzberger syndrome, hypodontia, incontinentia pigmenti*

Introduction

Incontinentia pigmenti (IP; OMIM#308300) is also known as Bloch–Sulzberger syndrome, Siemens–Bloch syndrome, melanoblastosis cutis linearis, or pigmented dermatosis. IP is an X-linked, genodermatosis, and multisystem disorder. The prevalence of IP is 0.7/100,000 (Orphanet Report Series, 2011) with mutations in the inhibitor of kappa B kinase gamma (IKBKG, Xq28; earlier known as NEMO). IKBKG gene encodes NFkB protein, which controls the expression of other genes involved in cell proliferation, immunity, and inflammation. IP predominantly affects females and is thought to be fatal for males *in utero*.[1,2] This paper provides a literature review and an IP case report of a 16-year-old female who presented to our pediatric dental clinic where we managed her dental concerns. This report has been cleared for publication by the School of Dental Medicine. Parents gave both written and verbal consent for the use of patient-related documentation and information for the purpose of publication and no identifiers are disclosed in accordance with HIPAA guidelines.

Case Report

A 16-year-old female was referred to our pediatric dental clinic by her primary care provider for a chief complaint of pain in the upper and lower left teeth while eating. The patient was 5'11" with a weight of 268l bs (body mass index 37.4, >99th percentile) and blood pressure of 106/60 mmHg. Her body temperature was 37.2°C and SpO_2 was 100%.

Reported history (birth, social, and developmental)

The patient was born in Peru (6.5 pounds) through normal vaginal delivery to a G_1P_1 woman. At birth, hydrocephaly, cephalomegaly, pigmentation, and blisters on the left side of her face-neck along with "wire-like" or "wooly" texture of hair were reported. The pigmentation appeared whorled following the Blaschko's lines (lines in the skin indicative of cell development, not seen in normal healthy individuals) and increased in intensity [light-colored to verrucous dark-colored, Figure 1a] as patient aged. No family history of similar findings was reported. The patient demonstrated cognitive and developmental delays (started walking at age 3). Patient's family moved to the US as undocumented

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immigrant and hence, any medicodental treatment would be done primarily on emergency basis (family had no financial support).

Neurologic findings

When the patient was 14 years of age, she presented to the neurology clinic subsequent to having a seizure. The patient was diagnosed with generalized tonic–clonic seizures with postictal headache and weakness and placed on the antiseizure medication (levetiracetam, 500 mg/day). The patient was found to be neurologically alert and responsive. At age 14, she had a head circumference of 69.8 cm before a ventriculoperitoneal shunt was placed, and after placement, head circumference reduced to 68.5 cm (normal range: 54–57 cm). She had an increased fullness on the left side of her face with normal facial muscle function [Figure 1].

A magnetic resonance imaging of her brain demonstrated the abnormalities with findings suggestive of left hemimegalencephaly. In addition, there was evidence of an Arnold–Chiari malformation I [cerebellar tonsils herniated 2 cm below foramen magnum, Figure 1c]. The maxilla appeared triangular in configuration and demonstrated linear defect (cleft) extending through the alveolar bone and hard palate between the maxillary right central and lateral incisor [Figure 1d].

Orthopedic findings

The patient had significant scoliosis since her early teenage years with an exaggerated curve in her cervical spine. She was wheelchair bound, and pes planus and genu valgum were noted by the orthopedists.

Orofacial findings

Facial extraoral examination revealed the left-sided hemihypertrophy with generalized hyperpigmented whorls along the Blaschko's lines exclusively on the left side [Figure 1a]. The posteroanterior skull view demonstrated maxillary and mandibular hemihypertrophy [Figure 1b]. Extraoral facial midline was not coincidental with intraoral midline [Figures 2b and 3a].

Intraoral soft-tissue examination revealed gingival hyperplasia with a high-arched palate with the left-sided hemihypertrophy of tongue. In addition, enlargement of the filiform papillae on the left side of tongue was noted [Figure 3b]. The patient had a history of traumatic fibroma due to chronic tongue biting while seizure.

Intraoral hard-tissue examination revealed that the generalized spacing between the teeth along with a midline shift of 10 mm to the right and the occlusal plane was accentuated [Figure 2a]. The size of the teeth was larger (wider) on the left side (~1.2X times) compared to corresponding teeth on the unaffected right side [Figure 2a-e]. Clinical examination revealed that the patient had an adult dentition except for missing third molars and #29 [Figure 2a-e]. Severe halitosis was reported by parents which was due to minimal tooth brushing by the patient.

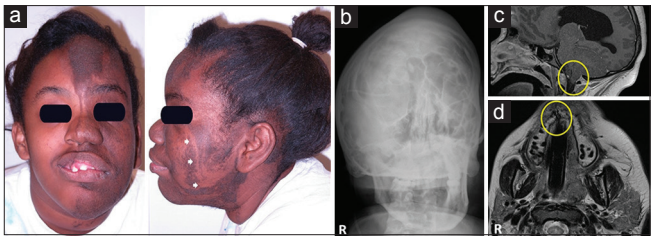

Figure 1: Frontal and left lateral facial view: left hemifacial hypertrophy with raised hyperpigmented patches along the Blaschko's lines indicated by arrows (a), posteroanterior radiograph of the skull: right side compensation (b), computed temography head images sagittal view: Herniation of cerebellum indicated in yellow circle (c) and axial view: gross left hemifacial hypertrophy and linear defect in maxilla between #7 and #8 area indicated in yellow circle (d)

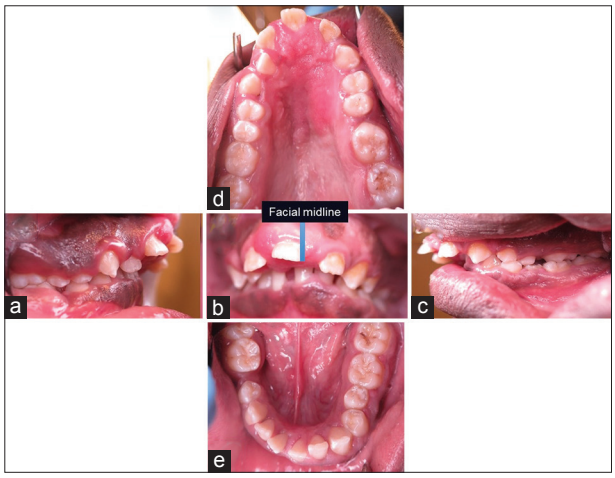

Figure 2: Intraoral images: right (a), frontal (b), and left (c) maxillary occlusal view (d) and mandibular occlusal view (e). The pictures demonstrate maxillary and mandibular hemihypertrophy with teeth on the affected left side appears to be larger in size than the unaffected right side. Midline deviation was evident (blue line indicating facial midline) with "gothic" or high-arched palate

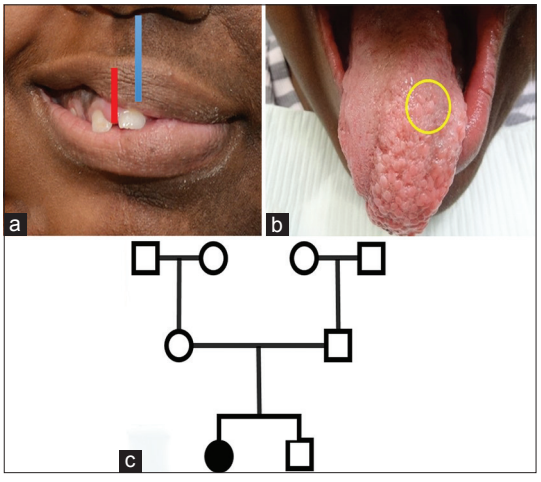

Figure 3: Smile line at rest with deviated midline to the right side due to the left-sided hemihypertrophy with blue line indicating facial midline and mesial border of #9 indicated by red line (a), hemihypertrophy of tongue on the left side with enlarged filiform papillae showed in yellow circle (b) pedigree analysis of three traceable generations suggestive of sporadic mutation in our patient (c)

Radiographic examination revealed the presence of generalized supra/subgingival calculus with no interproximal carious lesions. Due to patient's significant macrocephaly (and increased facial dimension) and cervical kyphosis, it was not possible to obtain a panorex that included both the sides of patient's face. Hence, it was necessary to take two images with different angle/focus for each side. The images were then superimposed to obtain a readable image. Impacted teeth #1, #17, and #32 were seen with a conical crown of #1. Tooth #29 appeared impacted and possibly resorbing the roots of #30 [Figure 4]. Occlusal caries were noted on #14, #15, #18, #19, and #20. The patient was determined as high caries risk based on risk factors including as follows: socioeconomic status, diet history, oral hygiene, medical history, and lack of a dental home.

Dental treatment

The patient never had a dental home. She was wheelchair bound and had a hard time sitting in the dental chair as any minor reclineation of the chair would trigger excruciating the neck and back pain. All dental care, including scaling and restorative dentistry, was completed with the patient seated in her wheelchair. The patient was appropriately referred to oral surgery for extractions and periodontics for subgingival scaling and root planning. Restorations were completed under local anesthesia and rubber dam isolation. The patient was not cooperative for restorative visits (F2: Negative behavior, Frankl rating). Despite our efforts to bring the patient back for follow-up care, she had only come for one follow-up appointment in subsequent 3 years. During this visit, we noted a new small carious occlusal pit, but improved oral hygiene.

Literature review

The condition was first described by A. E. Garrod in 1906.[3] It was named due to the histopathological characteristic and the presence of melanin pigment in the connective tissue cell layer, where it had been taken up by melanophores. Bloch (1925) and Sulzberger (1928) published cases with similar cutaneous manifestations.[3] IP manifestations include cutaneous lesions characterized by swirled hyperpigmentation along the Blaschko's lines with sparse, coarse hair. Cutaneous and noncutaneous manifestations of IP are summarized in Table 1.[1-15] Mendelsohn first described the orodental findings of IP[3] which included hypodontia, delayed eruption in both dentitions, and "peg-shaped" teeth.[9] A review of 40 IP cases revealed that approximately 75% of IP cases exhibited the presence of orodental findings.[1]

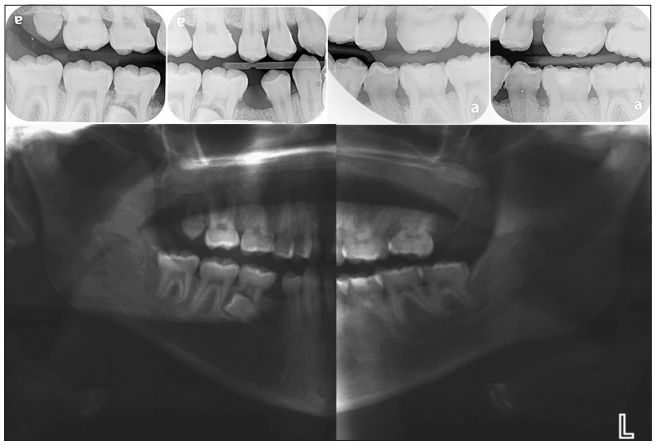

Figure 4: Bitewing and panorex radiographs. Panorex image is tailored form two different images from each side (see rationale in text). Bitewings and panorex show impacted #29 and resorption of #30 roots and impacted conical #1. Impacted #17 and #32. The left side permanent molars appear larger in size than the right permanent molars (radiographically the molars appears rotated, but clinically they are within normal limits)

Landy and Donnai in 1993 published diagnostic criteria for IP with emphasis on cutaneous findings as major criteria along with other findings as minor criteria.[4] Genetic basis for the development of IP has been extensively studied by Berlin *et al*.[7] *IKBKG* gene (2.8 kb, previously known as NEMO) is located on Xq28 and encodes a 419 amino acid protein, nuclear factor-kappa B (NFkB), or IKK-gamma. This NFkB (IKK-gamma) forms complex: IKK-alpha with IKK-beta. NFkB controls the expression of other genes involved in cell proliferation, immunity, and inflammation. In IP, this deletion of the 11.7 kb portion in the *IKBKG* (exon 4–10), leads to a downregulation of NFkB, which leads to increased apoptosis (cell death).[2,7] However, the *IKBKG* mutation was not added to the list of diagnostic criterion. IP is also characterized by some noncutaneous features. Therefore, in 2014, Minic *et al*. published a comprehensive analysis of additional diagnostic features of IP which appears to encompass the vast majority of all previously described features within this clinical condition.[8] Minic *et al*. expanded the use of cutaneous lesions as an IP major diagnostic criteria and suggested using dental, ocular, hair, nail, palate, breast, and central nervous system as minor diagnostic criteria [Table 2].[4,8] History of multiple male miscarriages, histopathologic features of IP, *IKBKG* mutation, and family history of similar condition could be used as additional criteria for IP diagnosis.[2] Targeted mutation analysis can be used to identify the most commonly occurring *IKBKG* deletion. If IP is suspected to be due to duplication or deletion of a less well-known portion of *IKBKG* gene, a skin biopsy should be considered. Histopathologic examination of biopsy sample would reveal hallmark features, melanin granules with large amount of eosinophilic infiltration in the dermis.[2] Once the diagnosis is made, genetic screening of all first degree relatives would prove helpful to rule out other affected individuals. A physical examination would provide more evidence for affected individuals. It is important to note that the patients can be diagnosed with IP regardless of failure to identify *IKBKG* gene deletion.[2] IP cases have been reported as a sporadic mutation due to the high mutation rate.[2] Kim *et al*. reported that unaffected parents might demonstrate germline mosaicism.[1] The hemifacial

Table 1: Cutaneous and noncutaneous clinical features of incontinentia pigmenti

Cutaneous clinical features and characteristics descriptions of lesions[1,3-5]	
Stages	**Characteristics**
Stage I	Description: Erythematous lesions leading to blisters
	Location: Whole body
	Duration: At birth-1 week and clears away by 4 months of age
	Hallmark: Linear distribution of lesion, leukocytosis, and marked eosinophilia
Stage II	Description: Verrucous or hyperkeratotic lesions
	Location: Distal ends of the extremities
	Duration: Starts after 4 months and heals by 6 months of age
	Hallmark: Warty lesions and rarely affects face and trunk
Stage III	Description: Hyperpigmented lesions ("marble cake" lesions)
	Location: Streaks of lesion on trunk (axilla and groin areas) and extremities
	Duration: After blisters are healed and remains by the second decade of life and then fades away
	Hallmark: Lesions follow the Blaschko's lines
Stage IV	Description: Hypopigmented lesions
	Location: Lower extremities and rarely trunk
	Duration: Can develop before or after hyperpigmented patches develop
	Hallmark: Alopecia, decreased vascularity, atrophic, and reticular nature
Noncutaneous clinical features[1,3,4,8-11]	
Dental (65%-75%)	Both dentitions affected
	Hypodontia
	Delayed eruption
	Tulip- or peg-shaped teeth
	Talon-cusp or accessory cusp
	Maxillary and/or mandibular hypoplasia
	Reduced vertical dimension
	Multiple impacted teeth (maxillary canine and mandibular teeth)
	Unilateral or bilateral cleft lip or palate
	Submucosal cleft
	Diathema or spacing
	Defective enamel and/or dentin mineralization (very rare)
Eye (60%-65%)	Squints
	Refractive errors
	Abnormal retinal vasculature (increased) with pigmented cells
	Retinopathy
	Cataract
	Blue sclera
	Nystagmus
	Strabismus
	Atrophy of optic nerve
	Microphthalmia
CNS (45%-50%)	Convulsive disorder or seizures (~66% of CNS manifestations)
	Intellectual disability
	Muscle spasticity or paralysis
	Global or developmental delay
	Mental or motor retardation
	Microcephaly
	Myelination delays
	Ventricular dilatation
	Agenesis of corpus callosum
	Polymicrogyria
	Periventricular leukomalacia
	Spina bifida
	Cerebral atrophy
Hair	Sparse and coarse structure ("woolly-hair nevus")
	Alopecia
	Sparse eyelashes and eyebrows
	Normal color of hair
Nails	If involved then majority of hand and feet nails are affected
	Spectrum of findings ranging from severity to mild form onychodystrophy to pitting/ridging (longitudinal)
	Painful subungual tumors
	Spoon-shaped finger nails
Breast	Breast hypoplasia with rudimentary or missing nipple
	Accessory or supernumerary nipples
	Abnormal nipple pigmentation (10 times higher than normal)
	Nipple asymmetry
Atypical features	Skeletal abnormalities
	Ear abnormalities
	Asymmetry
	Cleft lip and palate
	Scoliosis
	Leukocytosis (eosinophilia)
	Primary pulmonary hypertension
	Impaired fertility

CNS: Central nervous system

presentation of the condition in our case could suggest early chimerism (two different gene pools, one with IP and one without). Due to the hallmark clinical features in

Table 2: Diagnostic criteria

Criterion	No family history of IP	Family history of IP
Major criterion	Erythematous or vesicular neonatal rash	Skin lesions (including erythematous or vesicular rash, hyperpigmented, or hypopigmented lesions)
	Hyperpigmented lesions along Blaschko's lines	Dental abnormalities
	Linear hypopigmented hairless	Sparse-coarse hair
	IKBKG mutation*	Alopecia
		Ocular findings
		History of multiple miscarriages*
Minor criterion	Dental manifestations (hypodontia, tulip- or peg-shaped teeth, delayed eruption)	
	Ocular findings	
	Alopecia	
	Sparse-coarse hair	
	Pitting or ridging of nails	
	CNS abnormalities*	
	Cleft palate*	
	Nipple or breast abnormalities*	

IP: Incontinentia pigmenti, CNS: Central nervous system

Table 3: Conditions other than incontinentia pigmenti occurring along the Blaschko's lines

Condition	Features distinguishing from IP
NFJS	Reticular dermal pigmentation
	Heat intolerance
	Palmer hyperkeratosis
	Hypo-/hyper-hidrosis
	Alopecia
	Premature loss of teeth
	Enamel abnormalities
Hypomelanosis of Ito	Chromosomal mosaichism
	Cranial sutures abnormalities
	Hypopigmented dermal lesions
	Mental retardation
	Skeletal abnormality
	Asymmetry
	Deafness
	Scoliosis
	Disproportionate length of legs
	Hem hemimegalencephaly
	Abnormal head circumference
Chondrodysplasia punctate	Skeletal dysplasia
	Stenosis or thickening of cartilage
	Heart defects
	Cataract
	Alopecia with follicular pitting
	Commonly affects men

NFJS: Naegeli–Franceschetti–Jadassohn syndrome

this case, it would be valuable to have a genetic diagnosis. However, funding was not available for the laboratory tests (dental care was done through charity care provided by the School of Dental Medicine). Therefore, the diagnosis was based on the clinical features with the knowledge that the genetic diagnosis may have helped understand the etiology and pathophysiology of the mutation.

IP is thought to be primarily embryonically lethal in the male fetus and that reports suggesting that only 5% of male fetus survive.[1] Conditions such as Klinefelter syndrome (XXY) are a common genetic finding,[2] yet the finding of males surviving to birth with IP is not that common. Due to high rates of fetal demise for a male fetus, a history of multiple miscarriages is commonly reported in IP.[2] In the presented case, the mother reported no history of frequent miscarriages and the patient's older brother is medically healthy [See pedigree chart in Figure 3c]. Various studies documented variable ratios of affected females versus males 37:1,[11] 20:1,[1] and 14:1.[13] However, from one of the reports of 653 valid clinically identified IP cases, it was clear that IP affects individuals from all races.[16] There are no reports with documented differences in clinical findings between affected male versus female.[1-15]

Dental findings are observed in approximately 54%–75%[9] of IP cases. Commonly reported orodental findings are delayed eruption, hypodontia, and "peg-shaped" teeth affecting both primary and secondary dentitions.[1-15] Minic *et al*. reviewed articles published between 1993 and 2010 to systematically report orodental findings.[9] The group reviewed 1286 reported IP cases and concluded that the total number of dental and oral anomalies in each patient were more than 1 (1.67). The reported dental anomalies were shape deviations (36.4%), oligodontia (31.2%), and delayed eruption (17.8%). In addition, oral anomalies

included high arched ("gothic") or cleft palate (30%). Interestingly, surviving male IP patients have almost 50% chances of having high arched or cleft palate. One study also reported submucosal clefts.[13] Although some studies documented normal enamel and dentin structure in patients with IP,[3,9,15] one study reported enamel pitting or hypoplasia and hypocalcification.[13] Another study employed microanalytical tools to study the enamel structure and demonstrated that IP could significantly affect mineralization of enamel, but comparatively found no impact on the mineralization of dentin.[14] Uncommon reported dental findings include impacted maxillary canine and mandibular teeth,[13] maxillary and mandibular hypoplasia, or hemiatrophy.[9] Motamedi *et al*. reported the presence of talon cusp as one of the dental finding in IP.[10]

Here, the case presented demonstrated cutaneous, eye, hair, and dental findings. Majority of the findings were present on the left side of body. It is important to note that the most commonly reported dental findings such as hypodontia or oligodontia were not evident in our case. In addition, information regarding a delay in eruption of primary or permanent teeth could not be conclusively recalled by the patient's mother. The most commonly reported size variation was seen in our patient as well. It was manifested as "tulip-shaped" (comparatively larger crowns and smaller roots) teeth, but only on the affected left side. This type of hemifacial effect has not been reported previously. Teeth size variation between the affected and unaffected side was clearly evident in this case. Although minor variations in arch width and facial width on the right versus the left side have been documented in IP cases,[16] no reports documented hemifacial or tongue hypertrophy. In this reported case, we noted both hemifacial and tongue hypertrophy on the left side. Hemiatrophy of maxillary and mandibular arches has been reported in IP cases;[16] however, the substantial variations in arch widths, as evidenced in our case, have not been previously reported. We also noted additional orodental findings of IP such as "gothic" or high-arched palate, impacted mandibular teeth, palatal cleft (between #7 and #8), and diastema.

Cutaneous lesions in IP usually resolve by second or third decade of life. However, these lesions can be easily confused with other conditions especially if no family history of IP is known. Such conditions include epidermolysis bullosa, dermatitis herpetiformis, herpes zoster, bullous impetigo, and focal dermal hypoplasia. All these are distinct entities and histological examination can confirm IP diagnosis. X-linked hypohidrotic ectodermal dysplasia and immune deficiency and X-linked hypohidrotic ectodermal dysplasia with immune deficiency, osteoporosis, and lymphedema can be included in differential diagnosis of IP. However, both these conditions commonly occur in males, and NFkB function in these conditions may be impaired but not absent (as in IP).[2] In addition, some of the conditions which can follow the Blaschko's lines are presented in Table 3 along with the features that distinguish them from IP.[2] It is important to note that some of the findings in our patient match with that of Hypomelanosis of Ito (mosaicism characterized by hypopigmentation along the Blaschko's lines) such as scoliosis, hemimegalencephaly, and abnormal head circumference. However, hyperpigmentation confirmed the diagnosis of IP in our case. Furthermore, variable findings in IP are common, and additional gene mutations have been suspected to be present coincidentally for large phenotypic variation.[7]

Scheuerle and Ursini provided comprehensive information regarding management of IP affected individuals or children.[2] Multisystem involvement in IP would warrant a multidisciplinary management approach involves as follows: geneticist for counselling, pediatric dermatologist for management of cutaneous, hair and nail problems, pediatric neurologist for the management of seizures, pediatric ophthalmologist for the management of retinal hypervascularization or other ophthalmic problems, developmental pediatrician for the developmental or intellectual delay, pediatric dentist for the management of dental problems, and a craniofacial team for repair of clefts. It is important to note that none of the specialties have established guidelines for the management of patients affected with IP or other similar conditions. Regular follow-up is the only guideline mostly recommended by researchers and providers who frequently manage IP cases. Consistent with previous reports, our patient was managed by a team of developmental pediatrician, pediatric neurologist, pediatric dentist, periodontist, and oral surgeon. The patient positioning in the dental chair and panorex machine was a challenge due to the fact that the patient was obese and had Arnold–Chiari malformation I. Minor backward bending of the neck would cause excruciating pain to the patient. This could be a great concern for anesthesia and would possibly pose a significant problem for intubation. The patient was borderline cooperative, and hence, the most treatment was completed in her wheelchair with adequate neck support. Medical complexity warranted customized care for this patient. Due to family's immigration status, social, and financial issues, this patient was lost to follow-up. Multiple attempts were made to schedule the patient for follow-up; however, the limited family resources undermined the ability of the team to provide care.

There are still a few unanswered questions as to what is the role of NFkB in expression of full phenotypic features especially morphodifferentiation and eruption delay in IP. In addition, how IP features can be expressed on only one side of the body needs to be investigated. To understand this pathophysiology, mice deficient in *Nemo* (*IKBKG)* were generated using gene targeting. This mouse model could have served for understanding the phenotypic expression of IP in mice and extrapolating the information to humans for further research. However, the mice embryos could not survive past 13.5 days after gestation. The fatality was due

to tumor necrosis factor alpha-mediated apoptosis in liver and resulting liver damage.[17] In addition, female mice heterozygous for *Nemo (IKBKG)* were shown to have skin lesions similar to human lesion in IP.[18] Hence, the murine model can serve a valuable tool for future studies.

Conclusion

This is the first case of IP with hemifacial and tongue hypertrophy with tooth size and shape discrepancy between affected and unaffected side. Developmental and intellectual delay coupled with challenging social issues made the care and cooperation of this patient and family difficult and ultimately resulted in the patient being lost to follow-up. Hypertrophy of tongue and jaw has the potential to anatomically alter airway and inability to extend neck might pose challenges to the intubating patient for general anesthesia. Dental anomalies in IP cases may require esthetic or orthodontic corrections. Overall, a multiorgan problem needs a customized multidisciplinary care plan.

Important note

Further information can be found at the National Institute of Neurological Disorders and Stroke and IP International foundation websites which provide support for research and medical care of patients with IP.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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