Case Report

A Multidisciplinary Approach to a Seven Year-Old Patient with Incontinentia Pigmenti: A Case Report and Five-Year Follow Up

Rezvan Rafatjou [1], Fariborz Vafaee [2], Hanif Allahbakhshi [3], Porousha Mahjoub [4]

[1]Assistant Professor, Department of Pediatric Dentistry, Faculty of Dentistry, Hamadan University of Medical Sciences, Hamadan, Iran
[2]Associate Professor, Dental Research Center, Department of Prosthodontics, Faculty of Dentistry, Hamadan University of Medical Sciences, Hamadan, Iran
[3]Assistant Professor, Department of Prosthodontics, Faculty of Dentistry, Kashan University of Medical Sciences, Kashan, Iran
[4]Assistant Professor, Department of Pediatric Dentistry, Faculty of Dentistry, Guilan University of Medical Sciences, Rasht, Iran

Corresponding author:
P. Mahjoub, Department of Pediatric Dentistry, Faculty of Dentistry, Guilan University of Medical Sciences, Rasht, Iran

Porousha_65@yahoo.com

Received: 4 February 2016
Accepted: 12 June 2016

Abstract

Incontinentia pigmenti is a rare inherited disorder involving abnormalities of the skin, hair, eyes, musculoskeletal system, central nervous system, and the teeth. Dental abnormalities are the most common manifestations of this disorder. The purpose of this case report was to present the clinical and radiological findings of a seven-year-old girl as well as the results of her five-year follow up. The patient showed faded linear pigmented macular lesions on the trunk and on upper and lower limbs. Dental examination was notable for conical and peg-shaped anterior teeth as well as delayed eruption of primary and permanent teeth. In addition to conservative treatments, prosthetic treatments such as interim removable partial dentures were indicated for the patient.

Keywords: Incontinentia Pigmenti; Dental Prosthesis; Genetic Diseases, Inborn
Journal of Dentistry, Tehran University of Medical Sciences, Tehran, Iran (2016; Vol. 13, No. 4)

INTRODUCTION

Incontinentia pigmenti (IP) or Bloch-Sulzberger syndrome is a rare X-linked dominant disorder that is predominant among females. It is a systemic disease that involves tissues of ecto- and mesodermal origin, including the skin, teeth, eyes and the central nervous system, among other organs [1-4]. Incontinentia pigmenti affects 1 in 40,000 women and is usually lethal in men. It is secondary to mutations in the IKK-gamma gene, which is also known as NF-kappa-B essential modulator and is located on Xq28 [5,6]. NF-kappa-B essential modulator allows cells to respond to external signals such as growth factors and encodes a protein that controls the function of several chemokines, cytokines, and adhesion molecules, and is necessary for protection against tumor necrosis factor induced apoptosis [1,7,8].

Incontinentia pigmenti is characterized by erythematous eruptions with linear vesiculations, which are present at birth or appear soon after, and are mainly localized on the skin of the back, torso or the extremities [9]. Cutaneous eruptions along the lines of Blaschko are the hallmark of IP and develop in four different stages [10]:

1st Stage: Inflammatory, erythematous, vesiculobullous lesions, usually configured in a linear pattern (birth to one or two weeks);

2nd Stage: Papules and verrucous lesions with hyperkeratosis (two to six weeks);

3rd Stage: Hyperpigmentation of the skin (three to six months);

4th Stage: Hypopigmentation and atrophy of the skin (two to three decades).

Pigmentation gently fades and is usually absent by adulthood. Despite eosinophilia and leukocytosis on peripheral blood count, infants affected are generally not systemically compromised [11,12].

Prognosis of IP is generally good and depends on extra-cutaneous involvement, which may also affect patients' quality of life. According to the criteria presented by Landy and Donnai [13], skin lesions (erythema, hyper-pigmented streaks

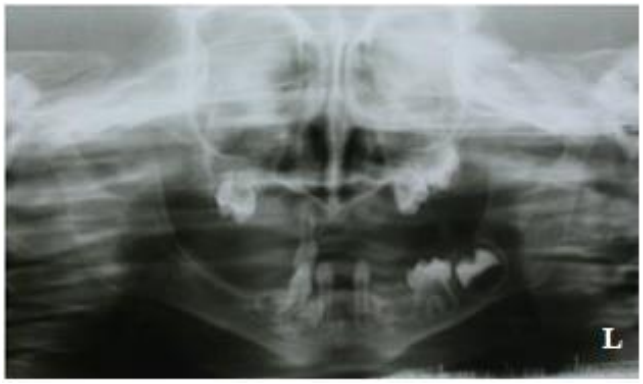

Fig. 1: Panoramic radiograph showing hypodontia and unerupted teeth at age two

and whorls, as well as pale, hairless, atrophic linear streaks or patches) were categorized as major diagnostic criteria for IP; whereas, dental, retinal, hair, and nail abnormalities are considered minor criteria for IP. According to the findings of some large case series of IP patients [2,8,14-17], the incidence of dental anomalies in patients with IP varies from 30.86% [14] to 92% [17]. Similarly, the number of dental and/or oral anomalies per patient ranges from 1.48 [8] to 2.48 [17]. Dental anomalies are the most common manifestations of IP patients and are observed in up to 80% of the patients and usually affect both dentitions. Hypodontia is the most common anomaly (up to 43% of patients), followed by pegged or conically crowned teeth (30% of patients) [18].In this report, we present a patient with IP who had multiple missing teeth in both the mandible and the maxilla, and provide a proper treatment plan for the patient's condition.

CASE REPORT

A two year-old Caucasian girl was referred to the Hamadan University of Medical Sciences, Faculty of Dentistry, for multidisciplinary evaluation and treatment of her dental condition. Her mother did not report any family history of IP and there was no consanguineous marriage. The child was born at full term after an uneventful pregnancy. She also reported the presence of a rash on upper limbs at birth that further developed into inflammatory vesicular alterations all over the body and cleared gradually, leaving linear pigmentations on the trunk and upper and lower limbs. Hematological tests at that time were normal. Physical and intellectual development were normal except for the skin findings that are outlined below. The patient was awake, alert, and cooperative.

The patient had woolly hair with normal distribution. Her skin was notable for linear pigmented macular lesions on the trunk, upper and lower limbs, and ears. She also had sparse eyebrows and eyelashes, xerophthalmia, and dry skin. Examination of the patient's oral cavity showed normal soft tissue with oligodontia and only four teeth were present (52, 72, 81, 82). There was no evidence of dental caries. Panoramic radiographs also showed oligodontia with eruption of left primary mandibular second molar (75), tooth germs of primary maxillary second molars (55, 65), left maxillary and mandibular permanent first molars (26, 36), mandibular permanent canines and right permanent lateral incisor (33, 43, 42) (Fig. 1).

Treatment plan at that time consisted of topical fluoride therapy with fluoride varnish every three months. The patient's family were given detailed instructions for oral hygiene. By the time the patient was seven years old she exhibited faded linear pigmented macular lesions on the trunk as well as the upper and lower limbs (Fig. 2). Dental examination was significant for conical and peg-shaped anterior teeth as well as delayed eruption of primary (52, 55, 65, 72, 75, 81, 82)

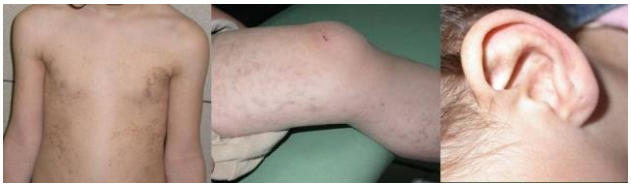

Fig. 2: Hyperpigmentation on the trunk, upper and lower limbs and ears

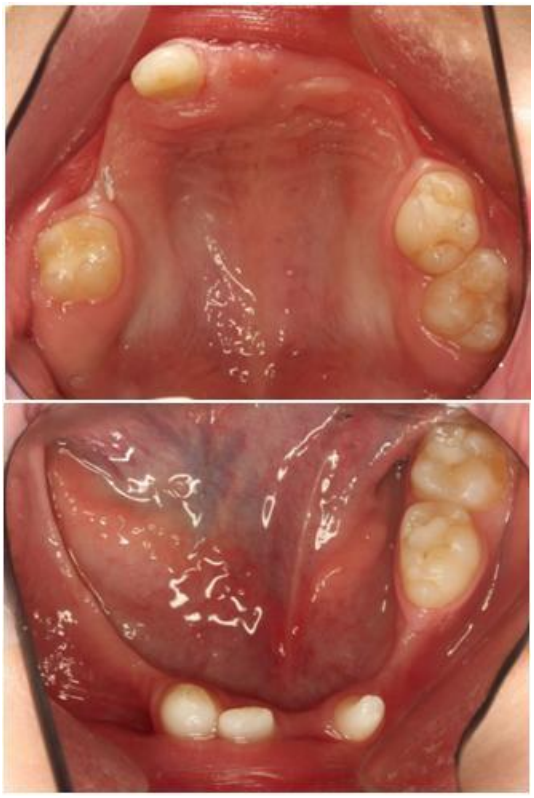

Fig. 3: Clinical view of the maxillary and mandibular arches at age seven

and permanent teeth (26, 36) (Fig. 3).

Panoramic radiographs revealed no dental or facial abnormalities except for multiple missing teeth. There were only six permanent tooth germs (Fig. 4) including number 12, which was malformed, and 25, 33, 37, 42 and 43. It was also found that tooth number 52 had dental caries.

The patient was initially treated with pulpectomy and composite filling of tooth 52, followed by fissure sealant therapy of teeth 55, 65, 75, 26 and 36 and composite veneering of teeth 52 and 82.

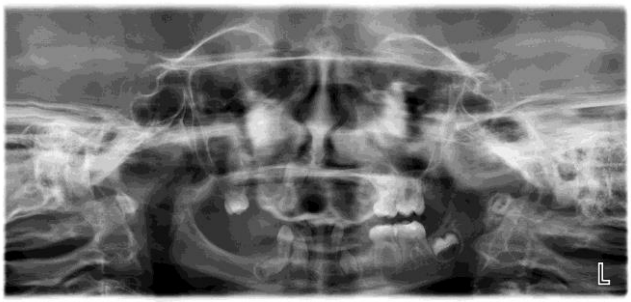

Fig. 4: Pretreatment panoramic radiograph; note the multiple missing teeth in both arches

In order to restore her masticatory function and improve her esthetics, removable partial dentures were fabricated for her according to the following instructions [19]:

1. Impression making for the study casts was performed with alginate material by using selected stock tray for children.
2. Special trays were made with acrylic resin.
3. Final impression making was performed with alginate (Alginoplast, Heraeus Kulzer, Wehrheim, Germany), using the trays and border molding with compound material.
4. Wax rims were made on the master casts.
5. Occlusal record was obtained with proper vertical dimension and freeway space was established.
6. A semi-adjustable articulator was used (A7plus, Bio-Art, Sao Carlos, Brazil).
7. Wax dentures were adjusted in the mouth.
8. The temporary dentures were made of heat-cured acrylic resin. At seven years and 10 months of age, the interim removable partial dentures were completed and inserted (Fig. 5). Figure 6 shows her frontal view after insertion of the dentures.

The patient and her mother were instructed that the dentures should be worn at all times except for while brushing and during sleep. The patient adapted to her dentures well. After a week, she complained of pain near the lingual flange of the lower denture. The denture was subsequently adjusted by checking the extension of the borders with light body silicone material (Speedex, Coltene, Alstatten, Switzerland). These multidisciplinary treatments were performed by a pedodontist and a prosthodontist.

DISCUSSION

Incontinentia pigmenti is an X-linked dominant disorder; therefore, the majority of the cases are seen in women [20]. In antenatal males, a mutant copy of the gene is inherited and can be lethal. It may rarely occur in men with Klinefelter syndrome (XXY syndrome) or as a result of

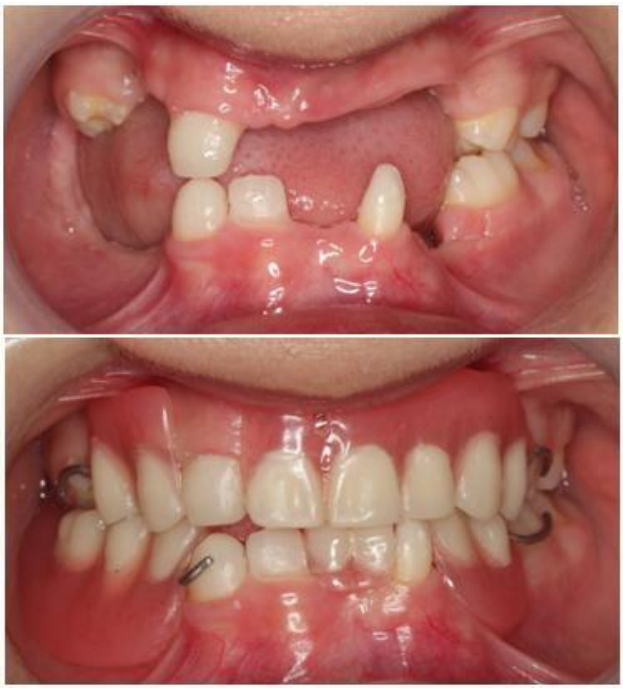

Fig. 5: Intraoral photograph before and after insertion of the interim removable partial dentures

somatic mosaicism or hypomorphic mutation in the NF-kappa-B essential modulator gene [18]. Incontinentia pigmenti is diagnosed in older children and adults by a series of skin manifestations, as well as possible dental, neurologic and eye abnormalities [21]. The clinical findings in this patient were consistent with those reported by others [11,12,22]. Her dental findings consisted of oligodontia, conical and peg-shaped anterior teeth and delayed eruption of primary and permanent teeth [22,23].

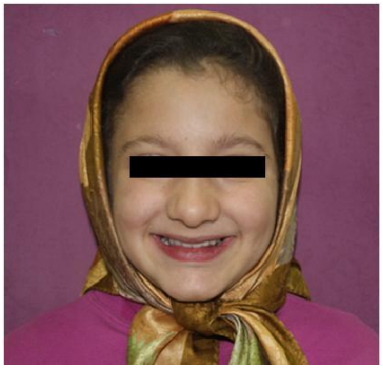

Fig. 6: Frontal smile view of the patient after inserting the interim removable partial dentures

Based on our findings and that of the other case series, it could be concluded that this triad is the characteristic dentition pattern of the patients with IP. There was no evidence of enamel defects, tooth malformation, missing teeth or ectodermic abnormalities in the other family members. Our patient only exhibited findings consistent with stage 1 (inflammatory or vesicular) and stage 3 (hyperpigmentation) of IP. Since the patient only suffered from xerophthalmia and dry skin, most of her complications were related to the stomatognathic system [1,3,24].

Some authors have reported subtle alterations of enamel and dentin structure that may evolve into caries, in addition to the typical dental findings of patients with IP [9,25]. Holmström et al, [16] reported decreased saliva secretion in 10 out of 25 IP patients, which caused dental caries, as saliva is protective against caries. Wang et al, [26] reported that different genes, or genes with differential effect sizes are involved in decay of primary and permanent dentitions. There were no other abnormal findings such as gothic plate that were reported by Minić et al, [15] and Himelhoch et al [27].

In spite of studies reporting hypocalcification and prevalent caries in patients with IP [27], our patient did not exhibit any enamel defects and dental caries were only detected in the primary maxillary lateral incisor.

Dental and oral anomalies may affect the patients' quality of life and cause feeding problems (hypodontia, cleft lip and cleft palate) but they are rarely life threatening [28]. Hypodontia can be treated by fixed or removable prostheses [29] or, in some cases, by tooth auto-transplantation [30]. To provide acceptable appearance for anterior teeth, composite was chosen for veneering and restoring carious teeth. Fissure sealant therapy is also recommended for non-carious, unrestored molars and premolars.

We prepared interim removable partial dentures to treat her masticatory problems and improve

her occlusal function and esthetic appearance. As the patient grows, her dentures will have to be modified and replaced. When the patient's growth spurt is completed, the removable prosthesis may be replaced by a permanent restoration such as osseointegrated implants. Implant-supported removable dentures also provide good stability and retention [31-33], but the placement of dental implants in young children is accompanied by several complications.

Yamashiro et al, [34] used orthodontic treatment to reposition the teeth in a 21 year-old female who had multiple missing and malformed teeth because prosthetic reconstruction without excessive tooth preparation was deemed difficult. Ocular defects including strabismus, cataract, conjunctival pigmentosa uveitis, optic atrophy, retinal vascular abnormalities, blue sclera, and microphthalmia are found in about 40% of the patients with PI. Retinal lesions may affect the peripheral retina and appear to be the result of ischemia with subsequent compensatory vaso-proliferation [18,35]. While central nervous system disorders such as seizures, mental retardation, spasticity, cerebral atrophy, hemiparesis, and encephalopathy occur in about 25% of the cases [36], our patient did not have any central nervous system abnormalities.

CONCLUSION

Since IP affects different organ systems to varying degrees, as seen in the present case, a multidisciplinary approach is recommended to treat and rehabilitate such patients. Patients suffering from IP should be constantly monitored by the ophthalmologists, oral surgeons and neurologists. As the patients grow up, periodic dental follow-ups should be scheduled to evaluate caries formation and the need for prosthetic reconstruction.

Patients with IP, like all other patients with hypodontia, require care from pediatric dentists, prosthodontists and orthodontists to achieve best results.

REFERENCES

1- Berlin AL, Paller AS, Chan LS. Incontinentia pigmenti: a review and update on the molecular basis of pathophysiology. J Am Acad Dermatol. 2002 Aug;47(2):169-87.

2- Hadj-Rabia S, Froidevaux D, Bodak N, Hamel-Teillac D, Smahi A, Touil Y, et al. Clinical study of 40 cases of incontinentia pigmenti. Arch Dermatol. 2003 Sep;139(9):1163-70.

3- Niccoli-Filho WD, da Rocha JC, Di Nicol R, Seraidarian PI. Incontinentia pigmenti (Bloch-Sulzberger syndrome): a case report. J Clin Pediatr Dent. 1993 Summer;17(4):251-3.

4- Francis JS, Sybert VP. Incontinentia pigmenti. Semin Cutan Med Surg. 1997 Mar;16(1):54-60.

5- Smahi A, Courtois G, Vabres P, Yamaoka S, Heuertz S, Munnich A, et al. Genomic rearrangement in NEMO impairs NF-kappaB activation and is a cause of incontinentia pigmenti. The International Incontinentia Pigmenti (IP) Consortium. Nature. 2000 May 25;405(6785):466-72.

6- Scheuerle AE. Male cases of incontinentia pigmenti: case report and review. Am J Med Genet. 1998 May 18;77(3):201-18.

7- Arenas-Sordo ML, Vallejo-Vega B, Hernández-Zamora E, Gálvez-Rosas A, Montoya-Pérez LA. Incontinentia pigmenti (IP2): Familiar case report with affected men. Literature review. Med Oral Patol Oral Cir Bucal 2004 Dec;10:E122-9.

8- Phan TA, Wargon O, Turner AM. Incontinentia pigmenti case series: Clinical spectrum of incontinentia pigmenti in 53 female patients and their relatives. Clin Exp Dermatol. 2005 Sep;30(5):474-80.

9- Bentolila R, Rivera H, Sanchez-Quevedo MC. Incontinentia pigmenti: a case report. Pediatr Dent. 2006 Jan-Feb;28(1):54-7.

10- Rosińska-Więckowicz A, Czarnecka-Operacz M. Bloch-Sulzberger syndrome: a case report. Postep Derm Alergol 2012 Sep;29(5):390-4.

11- Carney RG, Carney RG Jr. Incontinentia

pigmenti. Arch Dermatol. 1970 Aug;102(2):157-62.
12- Carney RG. Incontinentia pigmenti. A world statistical analysis. Arch Dermatol. 1976 Apr; 112(4):535-42.
13- Landy SJ, Donnai D. Incontinentia pigmenti (Bloch–Sulzberger syndrome). J Med Genet. 1993 Jan;30(1):53-9.
14- Fusco F, Bardaro T, Fimiani G, Mercadante V, Miano MG, Falco G, et al. Molecular analysis of the genetic defect in a large cohort of IP patients and identification of novel NEMO mutations interfering with NF-κB activation. Hum Mol Genet. 2004 Aug 15;13(16):1763-73.
15- Minić S, Novotny GE, Trpinac D, Obradović M. Clinical features of incontinentia pigmenti with emphasis on oral and dental abnormalities. Clin Oral Investig. 2006 Dec;10(4):343-7.
16- Holmström G, Bergendal B, Hallberg G, Marcus S, Hallén A, Dahl N. Incontinentia pigmenti. a rare disease with many symptoms. Lakartidningen. 2002 Mar 21;99(12):1345-50.
17- Hadj-Rabia S, Rimella A, Smahi A, Fraitag S, Hamel-Teillac D, Bonnefont JP, et al. Clinical and histologic features of incontinentia pigmenti in adults with nuclear factor-κB essential modulator gene mutations. J Am Acad Dermatol 2011 Mar;64(3):508-15.
18- Kitakawa D, Fontes PC, Magalhães FA, Almeida JD, Cabral LA. Incontinentia pigmenti presenting as hypodontia in a 3-year-old girl: a case report. J Med Case Rep. 2009 Nov 10;3:116.
19- Ohno K, Ohmori I. Anodontia with hypohidrotic ectodermal dysplasia in a young female: a case report. Pediatr Dent. 2000 Jan-Feb;22(1):49-52.
20- Shankar YU, Fatima N, Kumar MA, Prakash KS. Bloch Sulzberger syndrome (Incontinentia pigmenti): A rare case report with dental defects. J Indian Soc Pedod Prev Dent. 2015 Jan-Mar;33(1):74-7.
21- Garcia-Dorado J, de Unamuno P, Fernandez-Lopez E, Salazar Veloz J, Armijo M. Incontinentia pigmenti: XXY male with a family history. Clin Genet. 1990 Aug;38(2):128-38.
22- Gorlin RJ, Anderson JA. The characteristic dentition of incontinentia pigmenti. J Pediatr. 1960 Jul;57:78-85.
23- Cho SY, Lee AC, Drummond BK. Surviving male with incontinentia pigmenti: A Incontinentia pigmenti: a case report. Int J Paediatr Dent. 2004 Jan;14(1):69-72.
24- Dominguez-Reyes A, Aznar-Martin T, Cabrera-Suarea E. General and dental characteristics of Bloch-Sulzberger syndrome. Review of literature and presentation of a case report. Med Oral. 2002 Jul-Oct;7(4):293-7.
25- Macey-Dare LV, Goodman JR. Incontinentia pigmenti: seven cases with dental manifestations. Int J Paediatr Dent. 1999 Dec;9(4):293-7.
26- Wang X, Shaffer JR, Weyant RJ, Cuenco KT, DeSensi RS, Crout R, et al. Genes and their effects on dental caries may differ between primary and permanent dentitions. Caries Res. 2010;44(3):277-84.
27- Himelhoch DA, Scott BJ, Olsen RA. Dental defects in incontinentia pigmenti: case report. Pediatr Dent. 1987 Sep;9(3):236-9.
28- Minić S, Trpinac D, Gabriel H, Gencik M, Obradovic M. Dental and oral anomalies in incontinentia pigmenti: a systematic review. Clin Oral Investig. 2013 Jan;17(1):1-8.
29- Jocbsen P. Diagnosis and treatment planning for fixed and removable prosthodontics in Restorative dentistry. 2nd ed., Oxford, Wiley-Blackwell, 2008:154-5.
30- Jensen JL, Album B. Tooth transplantation in cherubism and incontinentia pigmenti: report of two unusual cases. Oral Surg 2011 Feb;4(1):39-43.
31- Guckes AD, Brahim JS, McCarthy GR, Rudy SF, Cooper LF. Using endosseous dental implants for patients with ectodermal dysplasia. J Am Dent Assoc. 1991 Oct;122(10):59-62.
32- Guckes AD, McCarthy GR, Brahim J. Use of endosseous implants in a 3-year-old child with ectodermal dysplasia: case report and 5-year follow-up. Pediatr Dent. 1997 May-Jun;19(4):282-5.

33- Kraut RA. Dental implants for children: creating smiles for children without teeth. Pract Periodontics Aesthet Dent. 1996 Nov-Dec;8(9):909-13.
34- Yamashiro T, Nakagawa K, Takada K. Case report: orthodontic treatment of dental problems in incontinentia pigmenti. Angle Orthod. 1998 Jun;68(3):281-4.
35- Chan YC, Happle R, Giam YC. Whorled scarring alopecia: a rare phenomenon in incontinentia pigmenti. J Am Acad Dermatol. 2003 Nov; 49(5):929-31.
36- Burton RH, Ebling JL, Rook FJG. Rook/Wilkinson/Ebling Textbook of Dermatology. 5th ed., Oxford, Wiley-Blackwell,1992:1580-2.