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Congenital Atrichia: A Case Report

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ABSTRACT

Introduction: Congenital Atrichia is a rare genetic condition in which all body hair may be absent. Congenital Atrichia is a rare genodermatoses is characterized by a mutation of the human hairless (HR) gene on chromosome 8p22. There is loss of scalp hair between, eyebrow, eyelash, and body hair may also be sparse or absent; patients may have a few pubic and axillary hairs. The condition may present in isolation or along with other defects. There are many forms of inherited alopecia (i.e., hair loss), which vary in age of onset, severity, and associated ectodermal abnormalities. Congenital alopecia universalis or congenital atrichia without associated ectodermal defects is a rare autosomal recessive disorder and is the only form of inherited alopecia for which the molecular basis is known. (2)

Case Report: Here is a case report on a preterm neonate delivered via normal vaginal delivery in our hospital setup. Patient was on mechanical ventilator after birth. Patient gradually improved at it was noticed that there was total absence of hair on the body. Dermatology opinion was taken and after few investigations and genetic testing it was confirmed case of Congenital Atrichia due to mutation in chromosome no 8p22.

Discussion: Congenital atrichia is a rare disease, causing irreversible alopecia which is inherited autosomal recessively. There is complete irreversible hair loss seen soon after birth. Situs inversus and mesocardia has also been reported to be associated with this disease.

Conclusion: Congenital atrichias represent a large and heterogeneous group of inherited hair disorders. In this report, we describe a patient affected with alopecia universalis congenita

Keywords: Congenital, atrichia, hairless gene, vitamin d deficiency

INTRODUCTION

Atrichia congenita (isolated form) is a rare autosomal recessive form of alopecia. Patients are born with hair that falls and is never replaced. The condition may be confused with congenital alopecia universalis, vitamin D dependant rickets, and ectodermal dysplasia. In our patients, congenital alopecia universalis was ruled out based on lack of any history of sudden, patchy loss of normally appearing hair progressing to the loss of scalp, body, eyelash, and eyebrow hair. Vitamin D dependent rickets was ruled out on the basis of lack of any history of joint pain, normal serum vitamin D3 and calcium levels, and a normal wrist joint radiograph. Ectodermal dysplasia was excluded as there was no history of delayed milestones or loss of sweating. Teeth, nails, mucosa (ectodermal components), palms, and soles were normal. The presence of a family history, history of consanguinity, and sparse eyebrows and eyelashes at the time of birth pointed to the diagnosis of atrichia congenita.

Zlotogorski *et al*. proposed diagnostic criteria for atrichia congenita with papular lesions but later Yip *et al*. revised them [Table 1].[1]

Major Criteria: Permanent and complete absence of scalp hair by the first few months of life Few to widespread smooth, whitish, or milia-like papules on the face, scalp, arms, elbows, thighs or knees from infancy or childhood Replacement of mature hair follicle structures by follicular cyst filled with cornified material in scalp histology Mutation (s) in the human hairless gene through genetic testing Clinical and/or molecular exclusion of Vitamin D dependant rickets
Minor (Supplementary Criteria) Family History of consanguinity Absence of secondary axillary, pubic or body hair growth and/or sparse eyebrows and eyelashes Normal growth and development, including normal bones, teeth, nails and sweating Whitish hypopigmented patches on scalp Lack of response to any treatment modality

Congenital atrichia is the complete absence of hair from birth. It can be inherited as an autosomal recessive or autosomal dominant or X linked pattern. The cases inherited as autosomal recessive are generally the most severe form and are present since birth. Congenital atrichia presents as total alopecia at birth, but sometimes scalp hair can be present at birth which is later shed between the first few months, after which no further regrowth occurs. There is alopecia of eyebrows, eyelashes, and general body hair.

Congenital atrichia may be associated with papular lesions which is a rare form of autosomal recessive syndrome characterized by numerous, small, horny papules on the face, neck, limbs, and trunk. Other syndromes associated with congenital alopecia are Moynahan's syndrome (mental retardation, epilepsy), Hidrotic ectodermal dysplasia (palmoplantar keratoderma, thickened nails) and aging syndromes. Skin histology shows the total absence of hair follicles or a few scattered miniaturized follicles. The gene for congenital alopecia has been mapped to chromosome 8p. Alopecia universalis and Vitamin D-dependent rickets type II A induced alopecia should be considered in the differential diagnosis.[2]

CASE REPORT

An extreme preterm (28 week) old male baby born to a $G_3P_2L_2$ mother via normal vaginal delivery, with birth weight of 900gms, did not cry after birth, was resuscitated according to neonate resuscitation protocol, was intubated in labour room with E.T. no. 2.5 and was shifted to NICU in view of no respiratory effort and was taken on ventilator with PEEP 5 cm H_20 and FiO_2 80%. On admission vitals HR- 154bpm, RR- 42/min, $Sp0_2$- 98% on ACMV mode of ventilator, CRT < 3 sec.

On General Examination: general condition of the patient was fair, HR – 152 bpm, RR - 40/min, SpO_2- 99% on ACMV mode of ventilation with FiO_2 80% and PEEP-5 cm H2O. Patient was started with ionotrope supports and IV fluids and prophylactic Antibiotic course and was kept NBM.

As patient-maintained oxygen saturation, FiO_2 was gradually decreased upto 30%. Extubation of patient was planned and patient was extubated, taken on CPAP mode of Ventilation with FiO_2 60% and PEEP 5cm H_20. As distress further reduced, FiO_2 was tapered down upto 30%. During the NICU stay, it was noticed that the baby had no hair on scalp, and had small white coloured mili like papules. Dermatology call was sent in view of the above-mentioned features and they suggested genetic testing of the baby. Trophic feeds were started slowly and feed was increased as the baby tolerated feeds well.

O_2 trial was given but the baby could not maintain saturation on O_2 by nasal hence was taken back on CPAP. Patient was on CPAP for few more days and then O_2 trail was given again and patient was taken on O_2. Antibiotics were stepped up after 14 days of NICU stay. Patient was taking full feed via OGT; hence IV fluids were stopped. Ionotropes were also tapered gradually and stopped when pulses were back to normal. Weight monitoring was done on daily basis and Oromotor stimulation call was sent at 32 weeks of gestation.

Patient was shifted to oral feeds when he started accepting orally well, and was

shifted to Step-NICU for weight gain. Serum Vitamin D levels were checked to rule out congenital Vitamin D deficiency and relatives were counselled regarding Genetic testing of the baby for congenital baldness. When baby was stable, accepting oral feeds well with consecutive weight gain for 7 days, baby was discharged with oral supplements. Relatives were also counselled about regular follow up to check for normal and Genetic testing on discharge. Genetic testing was done after few followups and mutation in 8p22 chromosome was detected. Thus, this case was confirmed as a case of congenital atrichia on the basis of fulfilment of major criteria according to diagnostic criteria.

Figure 1

DISCUSSION

Congenital atrichia is a rare disease, causing irreversible alopecia which is inherited autosomal recessively. There is complete irreversible hair loss seen soon after birth. Situs inversus and mesocardia has also been reported to be associated with this disease. Other associations include Moynahan's syndrome (characterized by seizures with mental retardation), hidrotic ectodermal dysplasia (palmoplantar keratoderma, thickened nails) and aging syndromes. Loss of scalp hair is seen between one to six months of age, after which no growth occurs. There may also be sparse or absent hairs on the eyebrow, eyelash, and body. Differential diagnosis includes vitamin D dependant rickets, congenital alopecia universalis, and ectodermal dysplasia. IFAP (Ichthyosis follicularis with alopecia and photophobia) syndrome has also been reported in literature where patient presents with a triad of striking alopecia, photophobia and generalized cutaneous 'thorn-like' projections.[3]

Atrichia has also been reported to be associated with anomalies of the face, nails, cartilage, speech, respiratory tract, digits, and oral cavity. Renal involvement in the form of nephrotic syndrome and glucosuria has also been reported. Ocular involvement in the form of congenital cataracts, microphthalmia, keratitis, congenital nystagmus, cone-rod dysfunction, and high myopia has been reported. Episcleritis is the inflammation of the episclera (a thin, loose, highly vascular connective tissue layer between the conjunctive and sclera). It can be idiopathic, or it can be associated with connective tissue disorders like rheumatoid arthritis, scleroderma and systemic lupus erythematosus. Watson and Hayreh classified episcleritis into simple and nodular. Most patients have intermittent bouts of moderate or severe inflammation at intervals of 1–3 months. Symptoms include acute onset of redness, lacrimation, and photophobia. It commonly affects a single quadrant in one eye. Bilateral involvement suggests underlying systemic disease. Systemic non-steroidal anti-inflammatory drugs (NSAIDs) and topical steroids are used in its treatment.[4] Congenital atrichias represent a large and heterogeneous group of inherited hair disorders. In this report, we describe a patient affected with alopecia universalis congenita (MIM 203655). Sequence analysis revealed a G to A transition at cDNA position 3034 of the hairless *hr* gene present in a homozygous state in the patient and in a heterozygous state in the patient's mother, and absent in the patient's sister. The mutation is predicted to result in the substitution of an asparagine residue for an aspartate amino acid (D1012N) at a position previously shown in the rat to affect hairless binding to thyroid hormone receptor. This study presents the first evidence in humans for the functional importance of the hairless thyroid

receptor interacting domain 2. (5)
Congenital alopecia (synonym: congenital atrichia) comprises localized as well as complete absence of hair at or shortly after birth. Congenital alopecia may occur isolated or with associated defects. On the basis of such associations, several different syndromes featuring congenital alopecia can be distinguished (1–3). The rare isolated form of congenital alopecia has been reported in sporadic and familial cases. In familial cases inheritance is usually autosomal recessive (MIM 203655) (2 , 4–11), but families with autosomal dominant (12) or X-linked recessive inheritance (MIM 300042) (13) have also been reported (6).

CONCLUSION

The proteins encoded by the human, mouse, and rat hairless genes contain a single ZF domain with novel spacing of a conserved six-cysteine motif. The mutation in this family, R620Q, resides between the fourth and fifth cysteine residues in the six-cysteine ZF domain (fig. 6). The mutated arginine residue has been conserved during the past 90 million evolutionary years, among human, mouse, and rat, suggesting that it is of significant importance in the function of the ZF domain. The hairless-gene product is a putative transcription factor with a single ZF domain, which is highly expressed in the brain and the skin. It has recently been shown that the suppression of hairless-gene activity in hairless mice results in several basic integument abnormalities at the cellular level, including complete disintegration of the outer root sheath of the hair follicle, failure of upward movement of the dermal papilla and subsequent induction of a new hair, and disruption of the integrity of key functional tissue units in the hair follicle (Panteleyev et al. 1998). In humans, the hairless gene appears to function at the cellular transition from the natal to the first adult hair cycle, and, if compromised, hair growth completely ceases and a new hair is never induced, and the result is a complete form of inherited atrichia.

Declaration by Authors
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