

Rapp-Hodgkin syndrome: An ectodermal dysplasia involving the teeth, hair, nails, and palate

Report of a case and review of the literature

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Rapp-Hodgkin syndrome is a rare form of ectodermal dysplasia involving the hair, eyes, sweat glands, nails, teeth, and palate. The case of a white girl with the condition is presented. The differential diagnosis is discussed, and the eight previously reported cases are reviewed. Another (ninth) previously reported case is considered for inclusion in the group.

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In 1968 Rapp and Hodgkin¹ described a family affected by a syndrome who displayed characteristic features of ectodermal dysplasia and multiple defects. Since then relatively few other cases have been reported,²⁻⁵ and none has appeared in the dental literature.

The characteristic features of the syndrome were summarized by Witkop and associates⁶ in 1975: stiff, sparse hair with the appearance of steel wool; sparse eyebrows and lashes; cleft palate; absent uvula; hypodontia; hypoplastic enamel; absent lacrimal punctae; epiphora; ectropion; photophobia; hypoplastic dermatoglyphics; and decreased number of sweat glands.

Subsequently, Silengo and coworkers³ microscopically examined the hair of a patient with this

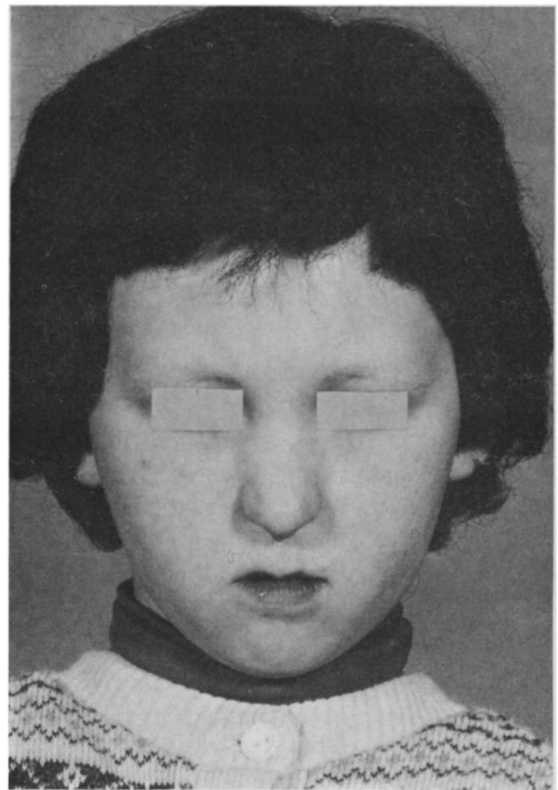


Fig. 1. Full-face view; the patient is wearing a wig.

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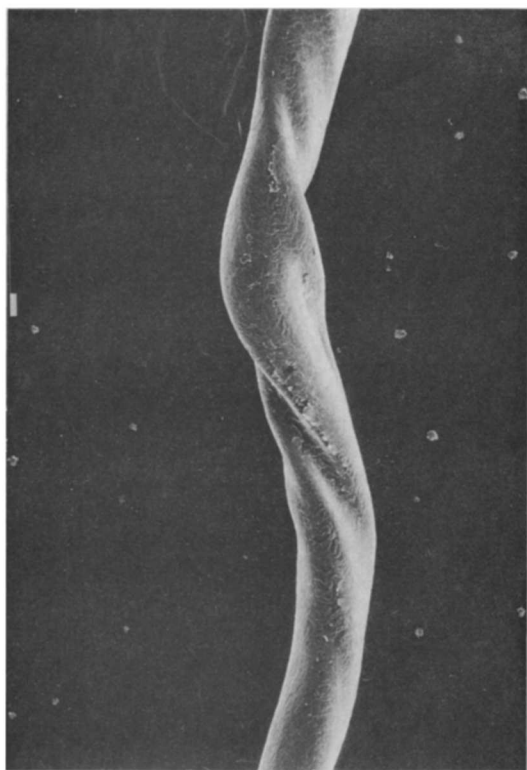


Fig. 2. Scanning electron micrograph of the patient's hair shows pili torti. (Original magnification, $\times 100$.)

condition. They reported the presence of pili torti (kinky hair).

CASE REPORT

The patient, a white girl, was first seen at age 4½ years for dental pain. Clinically (Fig. 1), she had a narrow nose with hypoplastic alae nasi, a small mouth, absent lacrimal punctae, epiphora, ectropion, photophobia, and partial deafness. Cutaneous syndactyly was not present. Her height and weight were above the 50th percentile. The patient's eyes showed a large amount of sclera, both laterally and below the iris. The scalp hair was coarse, kinky, and ginger colored, with a steel wool appearance; on microscopic examination revealed pili torti (Fig. 2). The eyelashes were sparse, and the lateral two thirds of the eyebrows were absent. All the toenails and fingernails were small and dysplastic and displayed soft tissue tufting (Fig. 3). Impressions of the finger pads were taken (Fig. 4) with polysulfide impression material as previously described.⁷ The epidermal ridges were irregularly flattened, with a reduced number of sweat pores (27/cm against a normal value of 36/cm at the same age⁷).

The patient's cleft of the soft palate that involved the uvula had been repaired. She was found to have the following deciduous teeth present, all of them grossly carious: right maxilla—two incisors, two molars; left maxilla—first incisor, two molars; right mandible—first



Fig. 3. Nail changes.



Fig. 4. Finger-pad impression shows irregularly flattened epidermal ridges and a reduced number of sweat pores.

incisor, canine, two molars; and left mandible—first incisor, canine, two molars.

In addition to the five teeth missing at first examination, a panoramic radiograph (Fig. 5) showed a number of permanent teeth to be congenitally absent, probably the maxillary right lateral incisor and second premolar, the maxillary left lateral incisor and second premolar, and both mandibular lateral incisors.

The crowns of the teeth as seen on the radiograph had an unusual appearance. The crowns of the first permanent molars and of the central incisors were rather square. Subsequent eruption of the first molars showed them to be hypoplastic, with a central occlusal depression rather than the usual fissure pattern. Each of the lower first permanent molars had three roots. The height of the coronal pulp chamber was increased relative to the overall length of the tooth. The upper first permanent molars had a taurodontic form (the pulpal portion was elongated at the expense of the radicular pulp).

The mandibular incisors, canines, and premolars exhib-



Fig. 5. Panoramic radiograph.

ited notching of the incisal edges, cusps, and occlusal surfaces, respectively (Figs. 6 and 7), a finding confirmed on eruption (Fig. 8). A similar notching of the maxillary premolars was suggested radiographically. The amount of enamel seemed reduced on all teeth, with a lack of contrast between enamel and dentin.

Cephalometric analysis, complicated by the absence of erupted teeth, suggested a class III dental base relationship. There appeared to be a maxillary retrusion rather than a mandibular protrusion. Sella-nasion (point A) was 76.5 degrees, sella-nasion (point B) was 75.4 degrees, and the maxillomandibular plane was 25.0 degrees. There was a tendency to an increased Frankfurt-mandibular plane angle (Fig. 9).

The mother stated that no other family members were affected, although the patient's sister has a degree of hearing impairment.

The patient reportedly had a small amount of pubic hair at 4½ years of age. During a recent episode of fever the patient had hallucinated and had required dousing with cold water. Under normal circumstances the patient reportedly tolerated heat quite well. A whole-back starch-and-iodine sweat test demonstrated an almost total absence of functioning sweat glands. Otitis media had been a recurrent problem.

REVIEW OF THE LITERATURE

Eight affected individuals in five families have been reported with this diagnosis.^{1-5, 8, 9}

Rapp and Hodgkin's¹ original family was composed of an affected mother and two affected children—a boy and a girl. Summitt and Hiatt² reported a similarly affected boy with the additional finding of hypospadias. Summitt and Hiatt were cautious, in light of this additional finding, not to align their case with those of Rapp and Hodgkin.

The original cases were further described by Wannarachue and coworkers,⁸ who noted the boy's

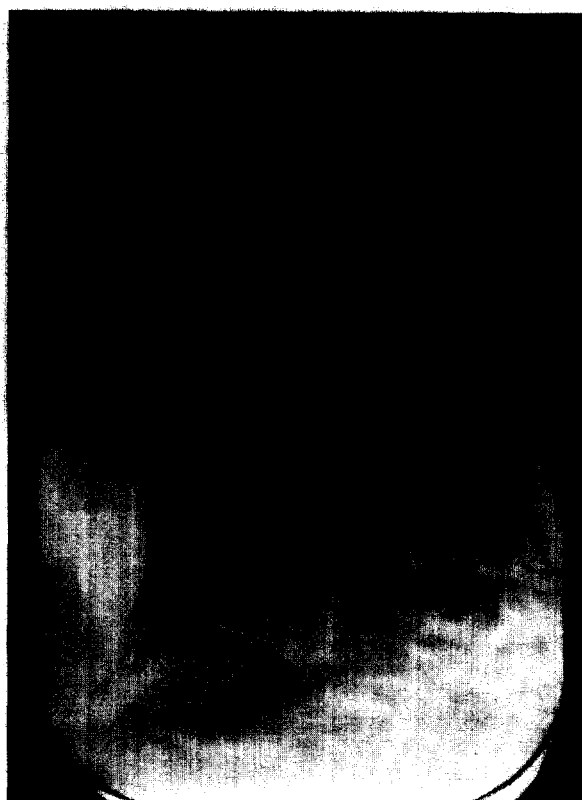


Fig. 6. Intraoral radiograph of lower incisor teeth.

hypospadias and thereby established this anomaly as part of the syndrome. Stasiowska's group⁹ reported an affected mother and daughter in whom Silengo and colleagues⁵ subsequently reported pili torti. This latter article also contained reference to personal communications from Schorr,³ who reported a further case in a female patient, and from Spaulding,⁴ who reported a similarly affected mother and her children. Beckerman¹⁰ reported the case of a female patient with anhidrotic ectodermal dysplasia and lacrimal anomalies. Although he did not record details of the child's facies and did not note whether the "coarse and dry hair" and "sparse brows and lashes" showed pili torti, in other respects the condition he described is similar to that described by Rapp and Hodgkin. The findings in these cases and the present case are compared in Table I.

The facies of Rapp-Hodgkin (RH) syndrome are somewhat characteristic (Fig. 1). This syndrome should be differentiated from a large number of ectodermal dysplasias with dental abnormalities. Of the four areas particularly afflicted—hair, nails, eyes, and palate—the two most effective "sieve" features are the pili torti and the cleft palate.

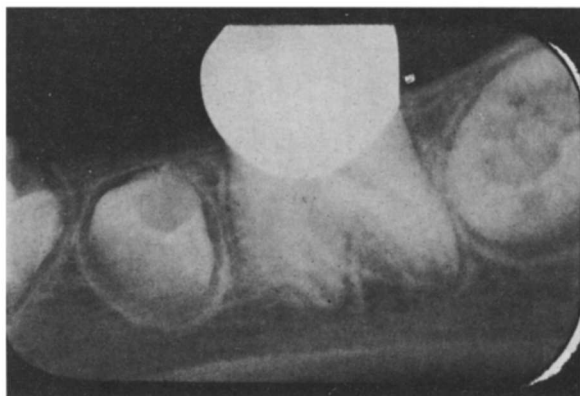


Fig. 7. Intraoral radiograph of lower left quadrant.

As recorded by Freire-Maia and Pinheiro,¹¹ there are seven forms of ectodermal dysplasia associated with pili torti^{1, 12-16} (Table II); of these, only RH syndrome shows clefting of the palate. Ectodermal dysplasia with syndactyly displays a high, arched palate but, in the presence of normal facies, has less severe eye involvement and spinal lordosis.

Freire-Maia and Pinheiro¹¹ recorded six diagnostic alternatives for patients with ectodermal dysplasia and cleft palate.^{1, 17-22} Many of these conditions show abnormal hair texture or distribution, and further diagnostic features must be taken into account (Table III). Of these other conditions, xeroderma-talipes-enamel defect¹⁷ includes bilateral clubfoot, which is not a feature of RH syndrome. Patients with Rosselli-Gulienetti syndrome (ectodermal dysplasia with cleft lip, cleft palate, and popliteal pterygium)¹⁸ bear the additional stigma of dystrophic facial skin that may exhibit desquamation and erythematous patches. These patients may also have papulofollicular dermatosis of the trunk with supernumerary nipples. Ectodermal dysplasia with ectrodactyly, cleft lip, and cleft palate¹⁹ is marked by palmo-plantar hyperkeratosis, eczematous and pigmented nevi, and invariable ectrodactyly, syndactyly, or clinodactyly. The syndrome involving ankyloblepharon, ectodermal defects, cleft lip, and cleft palate²⁰ also displays supernumerary nipples. Regional ectodermal dysplasia with total bilateral cleft,²¹ in which the reported patient was edentulous and had a complete bilateral cleft of lip, jaw, and palate and an aplastic premaxilla, differs from RH syndrome, in which the cleft is of the palate alone. The syndrome is comprised of cleft lip and palate, ectodermal dysplasia, hand and foot anomalies, and oligophrenia²² features an abnormal electroencephalogram,



Fig. 8. Intraoral view of notched incisors and restored permanent molars.

Dob: 18.1.79

Age: 7 Yrs 1month
FEMALE

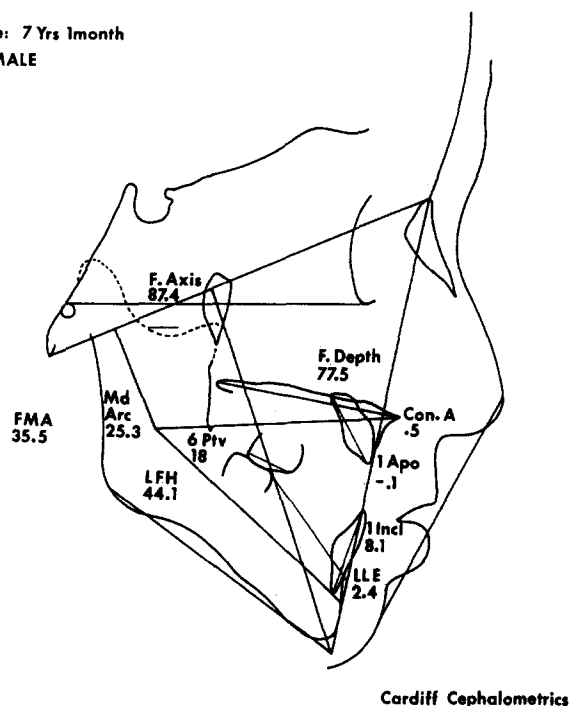


Fig. 9. Cephalometric tracing with Ricketts' analysis.

mental retardation, and syndactyly of toes and occasionally of fingers.

Not listed as an alternative by Freire-Maia and Pinheiro¹¹ is dysplasie ectodermique—division palatine—cheveux-chiendent (ectodermal dysplasia with cleft palate and "scrubbing-brush hair").²³ The affected individuals described in this report were a boy, his father, and two brothers. All had an overall marfanoid appearance, with coarse, matted hair

Table 1. Comparison of present case with previous cases

<i>Reference</i>	<i>Affected individuals</i>	<i>Hair</i>	<i>Teeth</i>	<i>Nails</i>	<i>Sweat</i>	<i>Skin</i>
Rapp and Hodgkin ¹ Wannarchue et al. ⁸	White ♀, Mother to: ♀ and ♂	Coarse, slow-growing scalp hair; sparse body hair; no pubic hair (but normal pubic hair in one case)	Hypodontia; conical teeth; incisors short and square	Dystrophic, distal soft tissue tufting	Decreased sweat production, variable (poor) response to heat; no sweat gland tubules or hair follicles on biopsy; infrequent palmar sweat pores	
Summitt and Hiatt ²	White ♂	Sparse, blonde, coarse, stiff "steel wool" hair; no axillary hair follicles on biopsy	All deciduous teeth extracted at 4 yr of age; possible enamel defect	Nails dysplastic	Sweats little; intolerant to heat	Dermatoglyphics hypoplastic and/or illegible
Schorr (cited in Silengo et al. ³)	♀	Blonde, wiry hair			Hypohydrosis	
Spaulding (cited in Silengo et al. ⁴)	♀, Mother and children					
Stasiowska et al. ⁹ Silengo et al. ⁵	♀, Mother to: ♀	Thick, reddish, curly hair; coarse, opaque, and wiry; pili torti; adult lost coarse, lustreless hair	Variable hypodontia; small conical teeth	Dysplastic, dystrophic	Sweating normal; infant sweats very little, with palmar sweat pores decreased in number	Dermatoglyphics: no ridges, very flattened ridges, hypoplastic dermal ridges
Crawford et al. (present case)	♀ White	Coarse kinky ginger "steel wool" hair; pili torti; small amount of pubic hair in childhood	Hypodontia, deciduous and permanent; square incisor forms; local hypoplastic defects	Dystrophic and tufted	One hallucination with fever; no back sweating with starch/iodine test	Irregularly flattened epidermal ridges
Beckerman ¹⁰	♀	Coarse, dry hair	Dental hypoplasia	Severe ungual dystrophy	No sweating	

showing a twisted, twin-electric-cable pattern under polarized light. Each had a repaired cleft palate, multiple dental defects, supernumerary nipples, nail defects, and some reduction in number of sweat

glands. The same authors describe a related and isolated case of a boy with similar but more severe stigmata: marked nail deformity; gross abnormalities of the form, number, and position of teeth; excess

<i>Hearing</i>	<i>Eyes</i>	<i>Face</i>	<i>General development</i>	<i>Limbs</i>	<i>Palate</i>	<i>Etiology</i>
Hearing problems secondary to otitis media	Eyelashes sparse; tarsal plate red; purulent conjunctivitis; eyebrows sparse, especially laterally	Midfrontal bossing, mildly depressed nasal bridge, high forehead	Mother 5 in shorter than her normal siblings; daughter, 50th percentile; son, 10th percentile; hypospadias		Unilateral cleft palate, absent uvula, short soft palate; cleft lip; bilateral cleft palate	New mutation in mother; ? autosomal dominant, possibly X-linked
Hearing normal; slitlike external auditory meatus	Eyelashes sparse, especially on lower lid; epiphora, ectropion; hyperemic lid margins; absent lacrimal punctae; corneal opacities; photophobia	Narrow nose, small mouth, hypoplastic maxilla, high nasal bridge	3rd Percentile; hypospadias		Bilateral cleft palate Cleft palate	
Otitis media-? associated with cleft palate	Eyelashes lost, epiphora; eyebrows sparse, lost laterally aplastic lacrimal canaliculi	Narrow nose, maxillary hypoplasia, small mouth, thin lips, high forehead	Normal intelligence; height; mother, 3rd percentile, but from a short family; daughter, 20th percentile, top limit of genetic potential	Partial cutaneous syndactyly, 2nd and 3rd toes, clinodactyly, 3rd toes	Cleft palate and bifid uvula	New mutation in mother; ? autosomal dominant possibly X-linked
Recurrent otitis media; partial deafness; 40-50 dB loss across speech range in both ears	Epiphora, ectropion, dyschiasis of eyelashes; absent lacrimal, punctae; no photophobia; reduced eyebrows	Narrow nose, hypoplastic alae nasi, small mouth	>50th Percentile; normal intelligence	No syndactyly	Repaired cleft of soft palate and uvula	Isolated case
Right-sided conductive hearing loss	Atresia of lacrimal punctae				Cleft palate with nasal speech	

sweating of hands, feet, and axillae with a characteristic strong odor. The areolae were present and normally pigmented, but the nipples were absent; there were no supernumerary nipples.

DISCUSSION

In this case report we have described a further case of the RH syndrome, the tenth reported case if that of Beckerman¹⁰ is included in the group.

Table II. Comparison of ectodermal dysplasia cases showing pili torti

	<i>Hair</i>	<i>Teeth</i>	<i>Nails</i>	<i>Sweat</i>	<i>Skin</i>	<i>Hearing</i>
Rapp-Hodgkin ¹	Coarse and stiff; pili torti; absent or scarce on scalp and body; sparse brows and lashes	Conical, short, square incisors or canines; hypoplastic enamel; caries and hypodontia	Small, narrow, dysplastic	Hypohidrosis; decreased number sweat glands	Dry and coarse; thick over extensor surface of knees and elbows; hypoplastic dermatoglyphics	Conductive loss 2° to otitis media
Trichoodontonychodysplasia with pili torti ¹²	Sparse, blonde scalp hair; pili torti	Hypodontia, widely spaced teeth, peg-shaped incisors; abnormal eruption	Dysplastic; flat, with linear grooves	Normal	Pale and mildly dry	Normal
Pili torti and enamel hypoplasia (Strandberg-Ronchese's dysplasia) ¹¹	Pili torti; monel-ethrix; thin, dry and blonde; if partial, affected hairs are lighter; bald areas; thin and twisted eyebrow hair	Enamel hypoplasia; spaced teeth; irregular shape; serrated biting edges; hypodontia; anodontia; delayed first dentition	Normal	Normal	Generalized keratosis pilaris	Normal
Pili torti and onychodysplasia (Beare type) ¹³	Normal in infancy; becomes hypotrichosis on the scalp; axilla and pubic hair coarse; pili torti	Normal	Short, fragile, brittle	Normal (1 case ? excessive)	½ Normal; ½ dry; ½ greasy	Normal
Salamon's syndrome ¹⁴	Dry, inelastic, wirelike, lustreless, sparse with telogen effluvium in the scalp; pili torti, trichorrhexis nodosa; scant brows, axillary, and pubic hair	Hypodontia; microdontia	Highly dystrophic, brittle, unguisplicatae; toenails more severely affected	Normal	Tendency to develop warts (verruca vulgaris) and papules (verruca plana); hyperkeratosis of the scalp	Not known

After Freire-Maia N, Pinheiro M. Ectodermal dysplasias: a clinical and genetic study. New York: Alan R. Liss, 1984.

As can be seen from Tables II and III, there are reports of many slightly different dysmorphic syndromes that may share features with RH syndrome but that are nonetheless distinct. A number of these conditions have seemingly unrelated features, such

as cleft hand,¹⁸ syndactyly,¹⁹ or cleft palate.¹⁷⁻²³ If each of these features is considered to arise from a defect of the ectoderm, such as a failure of an ectoderm-derived tissue-differentiation factor or a failure of programmed cell death, then closely related

<i>Eyes</i>	<i>Face</i>	<i>Development</i>	<i>Limbs</i>	<i>Palate</i>	<i>Other</i>	<i>Etiology</i>
Chronic epiphora; corneal opacities; photophobia; atresia of punctae, ectropion	Cleft lip; hypoplastic maxilla; mild frontal prominence; microstomia; mild depressed nasal bridge; prominent and malformed auricles	Short stature	Occasional syndactyly	Cleft, bifid uvula	Chronic rhinitis; nasal speech; hypospadias	Autosomal dominant? X-linked dominant?
Normal	Mild maxillary hypoplasia; thinness of lip	Normal	Normal	Not known	Not known	Autosomal dominant? X-linked dominant?
Chronic blepharitis	Normal	Normal	Normal	Not known	Not known	Autosomal dominant
Normal	Normal	Low IQ; severely retarded; irresponsible personality	Normal	Not known	Not known	Autosomal dominant
Blepharoconjunctivitis chronica; keratitis punctata; atrophica striata pigmenti retinae; madarosis and trichiasis palpebrae; maculae corneae centrales and periphericae	Pear-shaped nose	Normal	Slight long-bone osteoporosis	Not known	Not known	Autosomal recessive

pathways of causation might be postulated for the various findings in these ectodermal dysplasias. Conversely, it is possible that all these defects originate at the level of ectoderm-mesoderm interactions.

The present case again demonstrates the value of a multidisciplinary, cooperative approach to care.

Treatment in the present case has involved correction of the palatal defect, electrolysis for the aberrant eyelashes, and the provision of a wig. Dental extractions have been followed by the use of preformed stainless steel crowns for the intermediate restoration of the permanent molar teeth.

Table II. (cont.)

	<i>Hair</i>	<i>Teeth</i>	<i>Nails</i>	<i>Sweat</i>	<i>Skin</i>	<i>Hearing</i>
Arthrogryposis and ectodermal dysplasia ¹⁵	Hypotrichosis of scalp and body; scant brows and lashes; hair with nonkinky thin shaft is twisted about its own axis (stretched pili torti); oval cross section	Enamel hypoplasia	Absent at birth, later of normal length; tend to break longitudinally	Normal	Dry; tendency to excessive bruising and scarring after injuries and scratching	Normal; production of quantity of dry dark wax
Ectodermal dysplasia with syndactyly ¹⁶	Hypotrichosis; scalp hair is brittle, and either dark and thick or blonde and fine; pili torti; sparse brows and lashes	Severe crown hypoplasia; delayed and atypical eruption of permanent teeth	Yellowish and partly thickened	Normal	Dry with hyperkeratosis, especially over distal 1/3 of trunk, lower limbs, and palmo-plantar regions; transverse crease on both palms	Normal

Table III. Comparison of ectodermal dysplasias cases showing cleft palate

	<i>Hair</i>	<i>Teeth</i>	<i>Nails</i>	<i>Sweat</i>	<i>Skin</i>	<i>Hearing</i>
Xeroderma-talipes-enamel defect ¹⁷	Coarse and dry; slow growing; hypotrichosis; no lashes on lower lids	Poorly formed, yellow enamel	Deformed on the fingers and/or toes	Hypohidrosis; sweat glands small and few	Generally dry; scaling with ++ bullae on face and limbs; scant hair follicles	Not known
Rosselli-Gulienetti (Ectodermal dysplasia-cleft lip and palate-popliteal pterygia) syndrome ¹⁸	Woolly (negroid-type) thin, coarse, opaque, and short	Hypodontia; transverse striation; irregularities of the free margins	Subungual hyperkeratosis sulci; transverse and longitudinal striae; thinning of the lamina; irregularities of the free margins; hallucal nails with absence of the lamina	Palmar regions showed only a mild tendency to perspiration; not tested	Face dystrophic, tending to desquamation and erythematous patches; papulofollicular dermatosis on the trunk; supernumerary nipples; popliteal and perineal pterygium	Not known
Ectrodactyly-ectodermal dysplasia-cleft lip and palate syndrome ¹⁹	Hypotrichosis of scalp and body; fair and dry; scant or absent eyebrows and lashes	Anodontia; hypodontia; microdontia; enamel hypoplasia; poorly formed; increased caries; peg-shaped incisors	Dysplastic, thin, brittle, striated; pitted and terminated irregularly	Occasional hypohidrosis without hyperthermia	Dry, translucent, dystrophic; palmoplantar hyperkeratosis; eczematous patches; pigmented nevi	Conductive loss

<i>Eyes</i>	<i>Face</i>	<i>Development</i>	<i>Limbs</i>	<i>Palate</i>	<i>Other</i>	<i>Etiology</i>
Normal	Bilateral epicanthus; slight mongoloid slant	Short stature (<3rd percentile); probably low IQ	Bilateral clinodactyly; slight bilateral syndactyly of 2nd and 3rd toes; slight right talipes equinovarus	Not known	Arthrogryposis of all joints; diabetes mellitus; no menarche at age 16 yr	Not known

Mild crowding of lenses; discreet hypermetropia	Normal	Normal	Syndactyly of both fingers and toes, to variable degrees	High arched palate	Lordosis	Autosomal recessive
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<i>Eyes</i>	<i>Face</i>	<i>Development</i>	<i>Limbs</i>	<i>Palate</i>	<i>Other</i>	<i>Etiology</i>
Photophobia; hypoplastic ocular punctae; epiphora and blepharitis	Not known	Electroencephalographic alterations; mild mental retardation	Bilateral club foot	Cleft	Not known	Autosomal incomplete dominant
Not known	Cleft lip; hypoplastic auricular lobes; flat nasal pyramid with reduced subseptum	Not known	Aplasia or hypoplasia of thumb	Cleft	Malformed genitourinary system; absence or fusion of last lumbar vertebrae	Autosomal recessive

Tear duct anomaly or malfunction; speckled iris; photophobia; strabismus; blepharitis; clouding of cornea; congenital adhesions between eyelids	Cleft lip; broad nose; defective auricles (small, low set, reduced, amount of cartilage; posteriorly rotated); pointed chin, malar hypoplasia	Mental retardation	Ectrodactyly; syndactyly; clinodactyly	Cleft (not essential)	Renal abnormalities; rhinitis; respiratory infections; genital anomalies	Autosomal dominant
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Table III. Cont'd

	<i>Hair</i>	<i>Teeth</i>	<i>Nails</i>	<i>Sweat</i>	<i>Skin</i>	<i>Hearing</i>
Ankyloblepharon-ectodermal defects-cleft lip and palate syndrome ²⁰	Hypotrichosis (varying from dystrophic to sparse and brittle, almost totally absent scalp, axillary, and pubic hair); absent or scanty brows and lashes	Poorly formed and pointed; widely spaced; carious and discolored; severe hypodontia; delayed eruption	Severe dystrophy (variable from terminal dystrophy to total absence)	Hypohidrosis without hypothermia; decreased number of sweat pores	Dry and smooth; palmoplantar hyperkeratosis with obliteration of dermatoglyphic patterns; occasional reticulate hyperpigmentation; supernumerary nipples	Neural loss in 1 patient
Regional ectodermal dysplasia with total bilateral cleft ²¹	Scarce; present only on the skull circumference; eyebrows and lashes consist of short, thin, and very light-colored hairs	Anodontia	Normal	Anhidrosis on the head; defective sweat glands	Thin, shiny, desquamating on whole head, with dermoid cysts	No data
Rapp-Hodgkin	Coarse and stiff; pili torti; absent or scarce on scalp and body; sparse brows and lashes	Conical; short, square incisors or canines; hypoplastic enamel; caries+; hypodontia	Small, narrow, dysplastic	Hypohidrosis; decreased number of sweat glands	Dry and coarse; thick over extensor surface of knees and elbows; hypoplastic dermatoglyphics	Conductive loss 2° to otitis media
Cleft lip and palate, ectodermal dysplasia, hand-foot anomalies, and oligophrenia ²²	Scarring-type alopecia	Hypoplasia of enamel; missing teeth	Deformed nails; slow and irregular growth	Glands present but reduced in alopecic areas		
Dysplastic ectodermique-division palatine-cheveux-chien-dent ²³	Early scar appearance to center scalp; therein a gross epithelial thinning with no appendages; diffuse alopecia; black hair, sparse, dry, rough, with twisted form (pili torti); sparse lashes and brows (outer part lost)	Enamel "dysplasia"; malformed permanent maxillary right and left lateral incisors, absent mandibular right second molar	Onycholysis on some fingers (family); fingernails narrow and toenails hypoplastic, furrowed, thickened (single case)	No axillary sweating in response to heat; no axillary sweat glands; hands sweat satisfactorily; pores not assessed; probably apocrine absence	Family propositus has hyperelastic skin Single case—palmar and plantar hyperkeratosis; intact dermatoglyphics Family—supernumerary nipples Single case—absent nipple but areolae present	No data

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<i>Eyes</i>	<i>Face</i>	<i>Development</i>	<i>Limbs</i>	<i>Palate</i>	<i>Other</i>	<i>Etiology</i>
Lacrimal duct adhesion; photophobia; ankyloblepharon	Ankyloblepharon filiforme adnatum with partial fusion of eyelids at birth; broadened nasal bridge; hypoplastic maxilla; auricular abnormalities; cleft lip	Normal	Syndactyly	Cleft palate	1 case of genital and anal abnormality	Autosomal dominant
Ectropion; epidermalization of conjunctiva; hypoplastic tarsal plates; bilateral lagophthalmos in sleep	Bilateral cleft lip; aplasia of premaxilla	Normal	Normal	Bilateral cleft palate	Not known	Not known
Chronic epiphora; corneal opacities; photophobia; atresia of punctae; ectropion	Cleft lip; hypoplastic maxilla; mild frontal prominence; microstomia; mildly depressed nasal bridge; prominent and malformed auricles	Short stature	Occasional syndactyly	Cleft, bifid uvula	Chronic rhinitis; nasal speech; hypospadias	Autosomal dominant? X-linked dominant?
Photophobia; adhesions between eyelids		Abnormal electroencephalogram, mental retardation	Syndactyly of toes, occasionally also of fingers	Cleft lip, cleft palate	Chronic scalp infection; decubitus ulcer; blepharitis	
No data	Beak nose; hollowed nasal bridge; low-set ears; maxillary hypoplasia	Marfanoid appearance but normal stature; 1 case, mental retardation	Arachnodactyly; ligamentary laxity	Cleft palate	Single case—distinctive, strong odor to sweat	Family—autosomal dominant single case—Not known

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