

Frequent cutaneous manifestations of rare monogenic dental diseases: a review of OMIM data and cases from own clinical practice

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Abstract

In the last two decades, the elucidation of the genetic background of monogenic dental diseases has been significantly enhanced. In the Online Mendelian Inheritance in Man (OMIM) database there are 144 isolated or syndromic ones. Out of this 55 rare monogenic dental diseases (38%) are accompanied by cutaneous manifestations. In this study, we review the group of rare monogenic diseases with dental and cutaneous manifestations and observed the most frequent skin findings (hypohidrosis, hyperkeratosis, dry skin and skin fragility), nail symptoms (dysplastic nails and hypoplastic nails) and hair abnormalities (sparse hair, alopecia and hypertrichosis) and highlighted the genes associated with these frequent clinical features. We also summarized the frequent dental anomalies (missing teeth, abnormal shape of teeth, enamel abnormalities and delayed eruption or uneruption of teeth). Among the additional non-dental and non-cutaneous manifestations ophthalmological, skeletal and otorhinolaryngological abnormalities are the most frequently developing ones. Regarding the genetic background, there are 42 disease-causing genes associated with the 55 entities. Here, we also highlighted the *WNT10A*, *CTSC* and *EDA1* associated diseases in order to demonstrate how different variants of these genes can lead to the development of different phenotypes. Reviewing rare monogenic dental-cutaneous diseases, the association of the identified special clinical features may raise the attention of the specialist in everyday clinical practice and help in the identification of the underlying genetic background.

Key words: rare diseases, monogenic diseases, Mendelian, dental, cutaneous, manifestation



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Rare monogenic dental diseases with cutaneous manifestations

Intensive genetic and genomic research in recent decades has contributed tremendously to the elucidation of the genetic background of rare (prevalence 1:2000) monogenic diseases. In parallel with this, one of the consequences of today's digital development is that online available databases provide the widest range of insights in a given topic. These incredible developments are clearly demonstrated by the fact that in the Online Mendelian Inheritance in Man (OMIM, <https://www.omim.org/>) database 144 rare monogenic isolated

or syndromic dental diseases are presented. Among the rare monogenic dental diseases with extradental manifestations, there are 55 ones that are also associated with cutaneous manifestations.

In the case of suspected rare monogenic dental-cutaneous disease, detailed documentation of anamnestic data is essential to determine the exact clinical diagnosis (phenotype). Information must be obtained on dental abnormalities, cutaneous alterations and additional symptoms that may be associated. The inheritance of the disease within the family should be observed, drawing the family tree can provide significant help in clarifying the inheritance of the diseases.

Of the 55 rare monogenic dental-cutaneous diseases, 23 have autosomal dominant inheritance (AD; Table 1) and 27 entities are inherited with autosomal recessive manner (AR; Table 2).

Table 1. Rare monogenic dental-cutaneous diseases with autosomal dominant inheritance.

Disease	OMIM ID number	Disease causing gene	Anomalies				
			Skin	Nail	Hair	Teeth	Other
ACCES syndrome	619959	<i>UBA2</i>	✓	✓	✓	✓	✓
ADULT syndrome	103285	<i>TP63</i>	✓	✓	✓	✓	✓
Blepharocheilodontic syndrome 1	119580	<i>CDH1</i>		✓	✓	✓	✓
Cardioacrofacial dysplasia 1	619142	<i>PRKACA</i>		✓		✓	✓
Cardioacrofacial dysplasia 2	619143	<i>PRKACB</i>		✓		✓	✓
Ectodermal dysplasia 10A	129490	<i>EDAR</i>	✓	✓	✓	✓	
Ectodermal dysplasia 11A	614940	<i>EDARADD</i>	✓		✓	✓	
Ectodermal dysplasia 3	189500	<i>MSX1</i>		✓		✓	
Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3	604292	<i>TP63</i>	✓	✓	✓	✓	✓
Fontaine progeroid syndrome	612289	<i>SLC25A24</i>	✓	✓	✓	✓	✓
Genitopatellar syndrome	606170	<i>KAT6B</i>			✓	✓	✓
Glass syndrome	612313	<i>SATB2</i>		✓	✓	✓	✓
Hay-Wells syndrome	106260	<i>TP63</i>	✓	✓	✓	✓	✓
Lenz-Majewski hyperostotic dwarfism	151050	<i>PTDSS1</i>	✓		✓	✓	✓
Mandibulofacial dysostosis with alopecia	616367	<i>EDNRA</i>			✓	✓	✓
Naegeli-Franceschetti-Jadassohn syndrome	161000	<i>KRT14</i>	✓	✓		✓	
Oculodentodigital dysplasia	164200	<i>GJA1</i>	✓			✓	✓
Oligodontia-colorectal cancer syndrome	608615	<i>AXIN2</i>			✓	✓	✓
Rapp-Hodgkin syndrome	129400	<i>TP63</i>	✓	✓	✓	✓	✓
Scalp-ear-nipple syndrome	181270	<i>KCTD1</i>	✓	✓	✓	✓	✓
Singleton-Merten syndrome 1	182250	<i>IFIH1</i>	✓	✓		✓	✓
Tooth agenesis, selective, 4	150400	<i>WNT10A</i>	✓	✓	✓	✓	✓
Weyers acrofacial dysostosis	193530	<i>EVC2, EVC</i>		✓		✓	✓

Regarding the ratio of dominant and recessive syndromes, they occur in almost the same proportion among these diseases.

Among these rare monogenic diseases characterized by teeth and skin manifestations as well, there are only 5 with X-linked inheritance pattern (Table 3).

Molecular genetic testing is usually carried out after the identification of the clinical phenotype, with the written consent of the person concerned or his legal

Table 2. Rare monogenic dental-cutaneous diseases with autosomal recessive inheritance.

Disease	OMIM ID number	Disease causing gene	Anomalies				
			Skin	Nail	Hair	Teeth	Others
Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities	620519	<i>PPP1R13L</i>	✓	✓	✓	✓	✓
CDAGS syndrome	603116	<i>RNU12</i>	✓	✓	✓	✓	✓
Cranioectodermal dysplasia 1	218330	<i>IFT122</i>			✓	✓	✓
Cranioectodermal dysplasia 2	613610	<i>WDR35</i>			✓	✓	✓
Cranioectodermal dysplasia 4	614378	<i>WDR19</i>	✓			✓	✓
Craniosynostosis and dental anomalies	614188	<i>IL11RA</i>		✓		✓	✓
Dental anomalies and short stature	601216	<i>LTBP3</i>			✓	✓	✓
Ectodermal dysplasia 10B	224900	<i>EDAR</i>	✓		✓	✓	✓
Ectodermal dysplasia 11B	614941	<i>EDARADD</i>	✓		✓	✓	✓
Ectodermal dysplasia 16 (odontoonychodermal dysplasia)	257980	<i>WNT10A</i>	✓	✓	✓	✓	✓
Ectodermal dysplasia, ectrodactyly, and macular dystrophy	225280	<i>CDH3</i>			✓	✓	✓
Ehlers-Danlos syndrome, dermatosparaxis type	225410	<i>ADAMTS2</i>	✓		✓	✓	✓
Ellis-van Creveld syndrome	225500	<i>EVC, EVC2</i>		✓		✓	✓
Epidermolysis bullosa, junctional 1A, intermediate	226650	<i>LAMB3</i>	✓	✓	✓	✓	✓
Epidermolysis bullosa, junctional 1B, severe	226700	<i>LAMB3</i>	✓	✓	✓	✓	✓
Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	245660	<i>LAMA3</i>	✓	✓		✓	✓
Epidermolysis bullosa, junctional 4, intermediate	619787	<i>COL17A1</i>	✓	✓	✓	✓	✓
Epidermolysis bullosa, junctional 5A, intermediate	619816	<i>ITGB4</i>	✓	✓	✓	✓	✓
Epidermolysis bullosa, junctional 5B, with pyloric atresia	226730	<i>ITGB4</i>	✓	✓		✓	✓
Even-plus syndrome	616854	<i>HSPA9</i>	✓		✓	✓	✓
Haim-Munk szindróma	245010	<i>CTSC</i>	✓	✓		✓	✓
Hennekam lymphangiectasia-lymphedema syndrome 1	235510	<i>CCBE1</i>			✓	✓	✓
Oculodentodigital dysplasia	257850	<i>GJA1</i>			✓	✓	✓
Palmoplantar hyperkeratosis and true hermaphroditism	610644	<i>RSP01</i>	✓	✓		✓	✓
Papillon-Lefevre syndrome	245000	<i>CTSC</i>	✓			✓	✓
Schopf-Schulz-Passarge syndrome	224750	<i>WNT10A</i>	✓	✓	✓	✓	✓
Tooth agenesis, selective, 4	150400	<i>WNT10A</i>	✓	✓	✓	✓	✓

Table 3. Rare monogenic dental-cutaneous diseases with X-linked inheritance. XD: X-linked dominant, XR: X-linked recessive.

Disease	OMIM ID number	Disease causing gene	Mode of inheritance	Anomalies				
				Skin	Nail	Hair	Teeth	Other
Cornelia de Lange syndrome 5	300882	<i>HDAC8</i>	XD	✓		✓	✓	✓
Ectodermal dysplasia 1	305100	<i>EDA</i>	XR	✓	✓	✓	✓	✓
Kabuki syndrome 2	300867	<i>KDM6A</i>	XD			✓	✓	✓
Olmsted syndrome	300918	<i>MBTPS2</i>	XR	✓	✓	✓	✓	✓
Orofaciodigital syndrome I	311200	<i>OFD1</i>	XD	✓		✓	✓	✓

representative after genetic counseling. The task of clinical genetic counseling work is to provide information on the nature, etiology and risk of recurrence of the disease. In case of rare monogenic dental-cutaneous diseases, the specific, causal therapeutic modalities are not yet widely available, therefore the prevention and treatment of dental, cutaneous and additional non-dental and non-cutaneous manifestations is the primary goal of medical work.

Frequent skin abnormalities

Regarding the frequency of the different cutaneous manifestations of rare monogenic dental diseases, 37 diseases (67%) of the 55 are featured by the presence of skin manifestations. The observed skin symptoms in this group of diseases are the following ones: hypohidrosis, hyperkeratosis, dry skin, skin fragility, hyperhidrosis, and aplasia cutis congenita, sagging, redundant skin, photosensitive skin, dermatitis, wrinkled skin, keratosis pilaris (Fig. 1). Hypohidrosis and different forms of hyperkeratosis are the characteristics of 12 diseases (21%), dry skin is a common feature of 9 (16%) and skin fragility is a frequent symptom in 7 entities (12%).

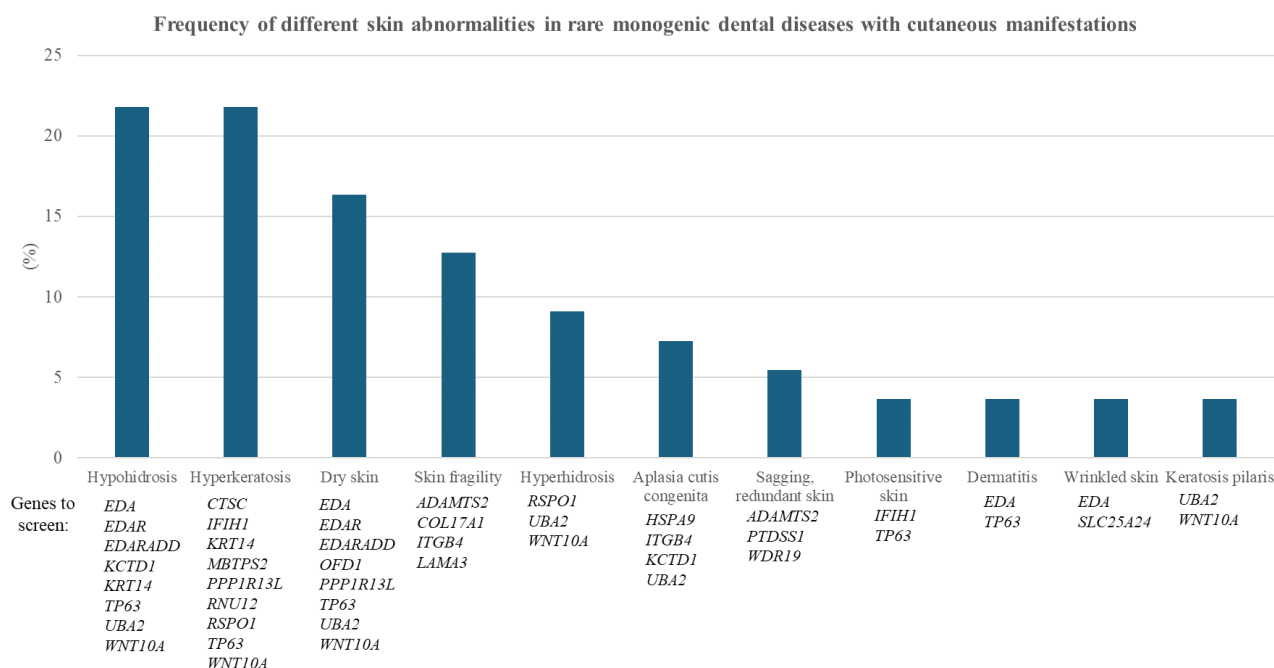


Figure 1. Skin abnormalities in rare monogenic dental diseases with cutaneous manifestations and disease-causing genes.

Frequent nail abnormalities

Regarding the frequency of the different cutaneous manifestations of rare monogenic dental diseases, 34 diseases of the 55 (61%) are featured by the presence of nail manifestations. The associated nail abnormalities are the following: dysplastic nails, nail ridging, nail pits, koilonychia, onycholysis and onychogryphosis (Fig. 2). Dysplastic nails are the characteristics of 25 diseases (45%) and hypoplastic nails are linked with 7 ones (12%).

Frequent hair abnormalities

Regarding the frequency of the different cutaneous manifestations of rare monogenic dental diseases, 40 diseases of 55 (72%) are featured by the presence of hair abnormalities. The associated hair symptoms are the following: sparse hair, alopecia, hypertrichosis, thin hair and wooly hair (Fig. 3). Sparse hair is a characteristic of 27 diseases (49%) and alopecia linked to 10 (18%).

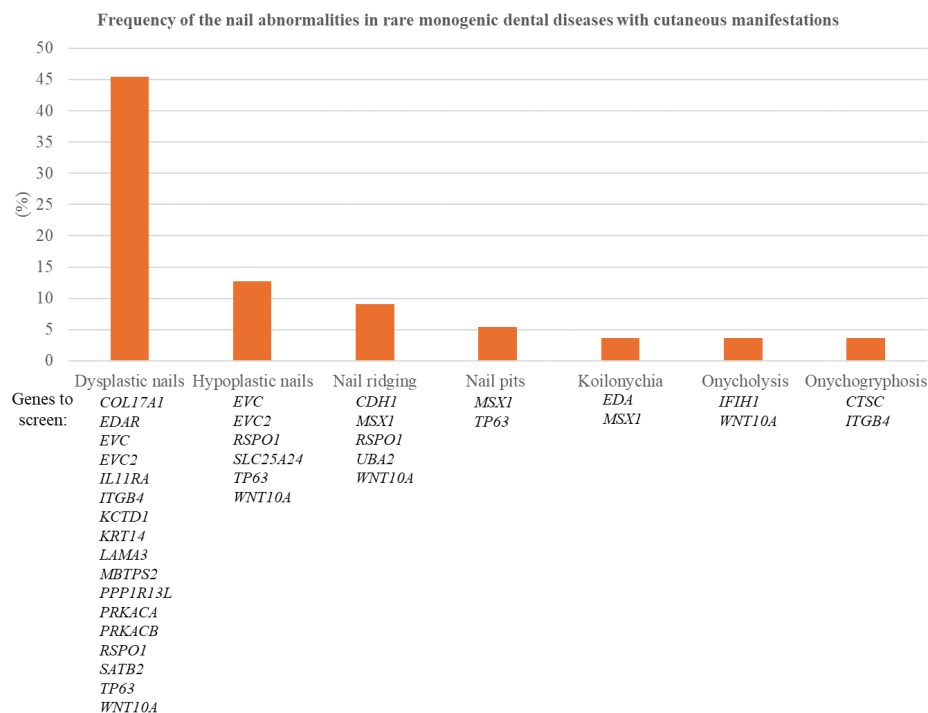


Figure 2. Nail abnormalities in rare monogenic dental diseases with cutaneous manifestations and disease-causing genes. The nomenclature of the nail symptoms are presented as they appear in the OMIM database.

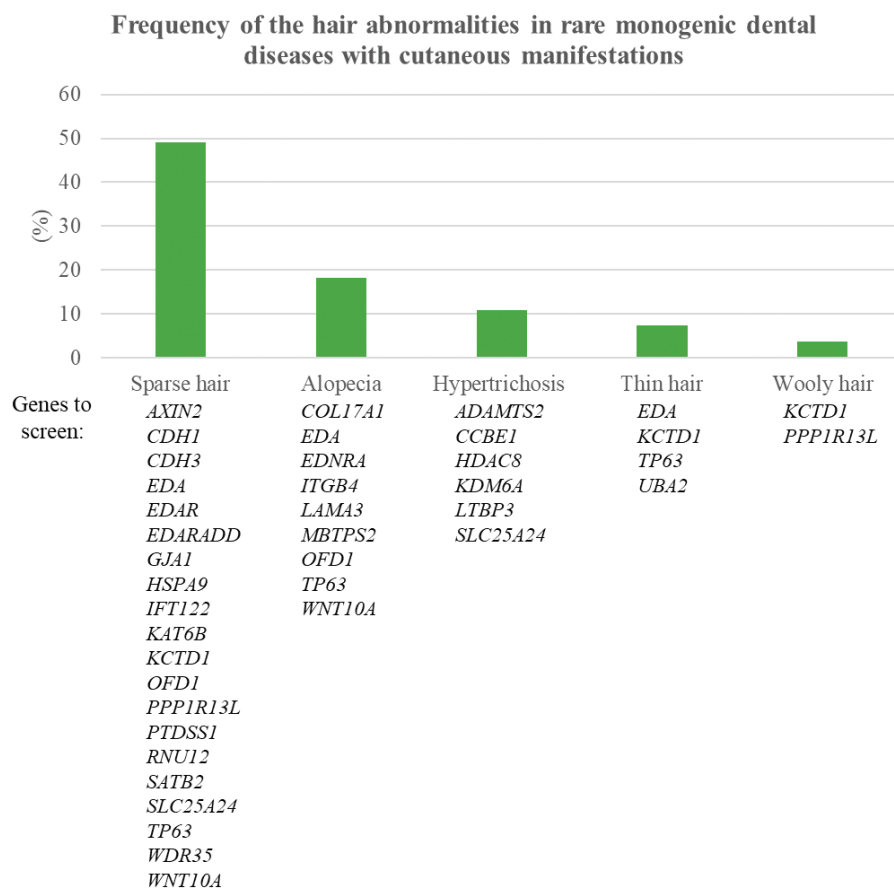


Figure 3. Hair abnormalities in rare monogenic dental diseases with cutaneous manifestations and the disease-causing genes.

Additional putative cutaneous manifestations

Besides the most common cutaneous manifestations in rare monogenic dental diseases, there are some clinical features, that are only associated with one specific disease among the reviewed rare monogenic dental diseases with cutaneous manifestations (Table 4).

Table 4. Additional putative cutaneous manifestations in rare monogenic dental diseases with cutaneous manifestations.

Disease	Additional putative cutaneous manifestations
ACCES syndrome	Adermatoglyphia, Frayed toenails
Naegeli-Franceschetti-Jadassohn syndrome	Reticulate hyperpigmentation, Absent fingerprints, Congenital malalignment of great toenails
Oculodentodigital dysplasia	Diffuse yellow-orange non-epidermolytic hyperkeratosis on palms and soles
Rapp-Hodgkin syndrome	Pili canaliculi
Singleton-Merten syndrome 1	Lentiginosities, Chilblain-like lesions, Subungual calcifications, Onycholysis
CDAGS syndrome	Porokeratosis, Paronychia, Downcurved nails
Schopf-Schulz-Passarge syndrome	Eyelid cysts
Cornelia de Lange syndrome 5	Naevus flammeus, Cutis marmorata

Frequently occurring teeth abnormalities in rare monogenic diseases with dental and cutaneous manifestations

It is important to know about the most frequent teeth abnormalities in the group of rare monogenic diseases with cutaneous and dental abnormalities, since it is easy to notice them during the dermatological examination. Missing teeth and abnormal-shaped teeth are the two most common features associated with monogenic dental-cutaneous diseases. Missing teeth are present in 35 (64%) of the investigated 55, while shape abnormalities of the teeth are a frequent symptom in 34 (62%) of this group. Enamel abnormalities are associated with 12 entities (22%), while delayed or unerupted teeth are linked with 11 diseases (20%). Other relatively rarely detected teeth abnormalities are hypernumerary teeth, chronic periodontitis and dentin abnormalities.

Among the other non-dental and non-cutaneous manifestations ophthalmological, skeletal, otorhinolaryngological, neurological, urogenital, pulmonary, cardiovascular and gastrointestinal abnormalities occur frequently, however, the most common ones are the ophthalmological and skeletal anomalies.

Variants of the same disease-associated gene can lead to the development of variable phenotypes and clinical entities

In the case of the presented pathologies, the exact description of the dental-cutaneous and additional possible clinical symptoms, as well as the clarification of the inheritance process, can greatly facilitate the establishment of the diagnosis. Molecular genetic testing can confirm the established clinical diagnosis by clarifying the genetic background. At the same time, during the everyday clinical genetics routine, we also encounter quite complex cases. In the following, by presenting some of our own cases, we highlight that different variants of the same gene can result in different clinical entities, moreover, even the

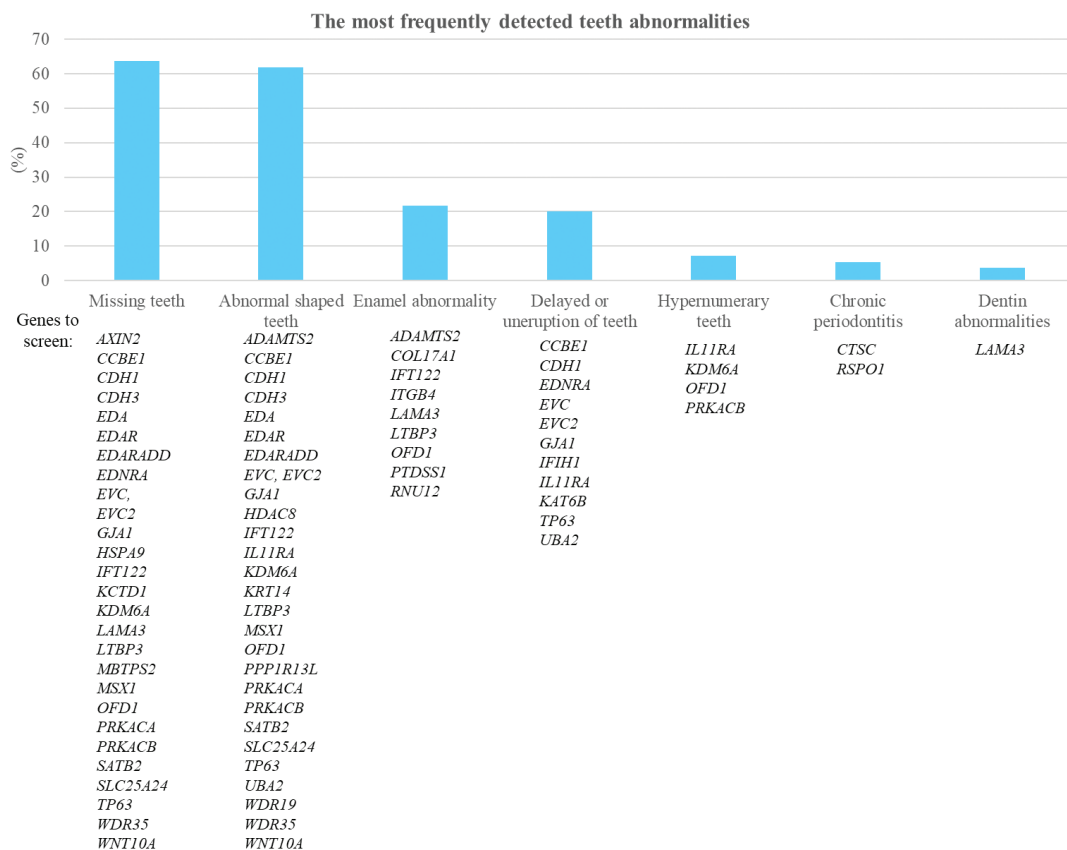


Figure 4. The most frequent teeth abnormalities in diseases with dental and cutaneous manifestations and disease-causing genes.

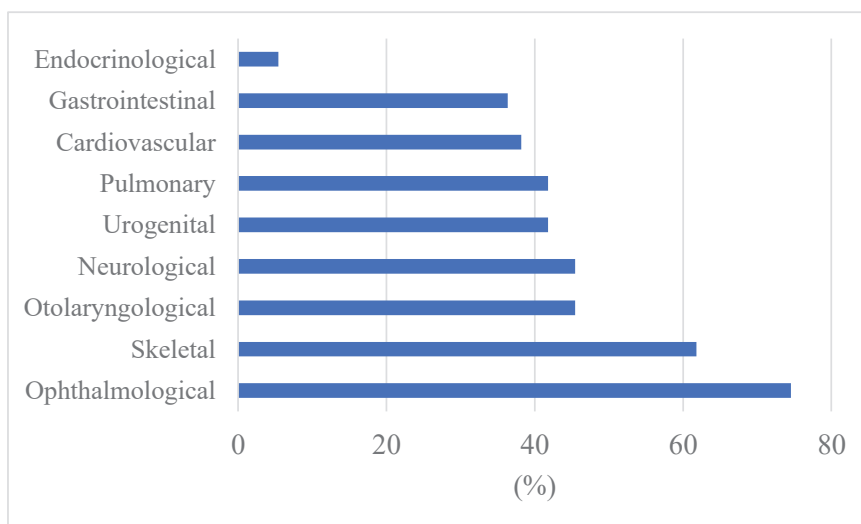


Figure 5. Additional organ manifestations among diseases with dental and cutaneous phenotypes.

same variants of the same gene can cause the development of different clinical symptoms in the affected patients. With our cases, we would like to draw the attention to the fact that the phenotypic variability can be quite large in the case of rare, genetically determined dental symptoms (also) associated with cutaneous symptoms, and how important it is to involve a clinical geneticist in clarifying each such case or family.

Regarding the genetic background, the rare pathogenic or likely pathogenic mutations of 42 disease-causing genes have been associated with the group of rare monogenic dental diseases with cutaneous manifestations. The explanation for this phenomenon is that different variants of the same disease-causing gene can lead to the development of different entities. As an example diseases caused by variants in the wingless-type MMTV integration site family member 10A (*WNT10A*) gene can be inherited as AD or AR and can lead to the development of 3 different rare diseases such as tooth agenesis 4 (STHAG4; OMIM 150400), odontoonychodermal dysplasia (OODD; OMIM 257980) and Schöpf-Schulz-Passarge syndrome (SSPS; OMIM 224750) [1–4]. These three entities have overlapping dental features such as the genesis of a variable number of permanent teeth (most commonly missing upper and lower premolars), small, pin-shaped teeth, and conical teeth. In OODD and SSPS, there may be palmar plantar hyperkeratosis and hypotrichosis including sparse eyebrows, sparse, dry, thin, brittle hair and nail abnormalities [1,2]. In the differential diagnosis of OODD and SSPS, the presence of eyelid cysts is decisive, in the case of which the SSPS clinical diagnosis arises [1,2].

In connection with the detection of a case in a British family, we confirmed that two different clinical variants associated with mutations in the *WNT10A* gene, such as OODD and SSPS, can even occur within the same family (Table 5). Eyelid cysts previously attributed to SSPS were present in a family member carrying the homozygous nonsense mutation (p.Cys107X) of the gene, while in homozygous missense (p.Phe228Ile) or compound heterozygous patients (1 heterozygous missense and 1 heterozygous nonsense mutation carriers) the OODD clinical variant was formed [1,2]. In the British family, dental symptoms were the most common among all clinical symptoms in the family [1,2].

Table 5. Clinical symptoms and genetic screening results of the British family affected by SSPS and OODD within the same family [2].

Patient ID No.	1.	2.	3.	4.	5.
Phenotype	SPSS	OODD	OODD	OODD	OODD
Eyelid cysts	+	-	-	-	-
Hyperkeratosis of the palms and soles	+	+	+	-	-
Hyperhidrosis	+	+	+	-	-
Dystrophic nails	+	+	-	-	-
Hypotrichosis	+	-	+	-	-
Microdontia	+	+	+	+	-
Oligodontia	+	-	-	+	-
Conical shaped teeth	+	-	+	+	-
p.Cys107Ter variant of the <i>WNT10A</i>	homozygote	heterozygote	heterozygote	heterozygote	-
p.Phe228Ile variant of the <i>WNT10A</i>	-	heterozygote	heterozygote	heterozygote	homozygote

Another interesting feature of our research is that the missense mutation we detected (p.Phe228Ile) was first identified in an US family affected by STHAG4, where hypodontia affecting the lateral incisors and premolar teeth developed in the patients [3]. Furthermore, Van den Boogaard et al. [4] detected the missense (p.Phe228Ile) and nonsense (p.Cys107X) mutations in 11 patients with tooth agenesis, who did not show the characteristics of OODD or SSPS in ei-

ther homozygous, heterozygous or compound heterozygous form [4]. All of this clearly illustrates how varied the clinical symptoms can be even in patients carry variants of the same gene or even exactly the same rare variant.

Entities caused by *CTSC* gene variants show an AR inheritance pattern and can result in the development of 3 different entities, whose common clinical feature is the development of aggressive periodontitis. In aggressive periodontitis type 1 (PPP; OMIM 170650) there are no associated extradental symptoms, while in Papillon-Lefevre syndrome (PLS; OMIM 245000) and Haim-Munk syndrome (HMS; OMIM 245010) the clinical picture is characterized by hyperkeratosis of the palms and soles. Other clinical characteristics of HMS are the development of pes planus, acro-osteolysis, arachnodactyly and nail abnormalities. In the case of a Hungarian female sibling pair from Szeged, dental and dermatological symptoms appeared in early childhood and the dermatological symptoms developed first. Due to the rapidly progressive, generalized periodontitis characteristic of the disease, early loss of milk teeth and later involvement and loss of permanent teeth appeared in both patients. Genetic screening identified a homozygous missense mutation (p.Gly301Ser) in the *CTSC* gene in both members of the examined sibling pair suffering from Papillon-Lefevre syndrome. The significance of our studies is that they draw attention to a rare disease, the Papillon-Lefevre syndrome, in which the first symptoms that develop can also be dental symptoms [5,6]. Moreover, investigating two independent Hungarian patients from Kaposvár, the genetic screening of the *CTSC* gene revealed the presence of the same homozygous nonsense mutation (p.Arg250X), however one patient exhibited the PLS phenotype and the other developed HMS (Fig. 6) phenotype [7]. Although these patients were not aware that they were related, haplotype analysis (especially genotypes of the rs217116 and rs217115 polymorphisms) clearly indicated that the patients carry the same haplotype, whereas unrelated healthy controls carried several different haplotypes.



Figure 6. Palmar hyperkeratosis, arachnodactyly and nail abnormalities of the HMS patient from Hungary.

Whole exome sequencing revealed two putative phenotype-modifying variants in these two patients: a missense mutation (rs34608771) of the SH2 domain containing 4A (*SH2D4A*) gene encoding an adaptor protein involved in intracellular signalling of cystatin F, a known inhibitor of the cathepsin protein, and a missense variant (rs55695858) of the odorant binding protein 2A (*OBP2A*) gene, influencing the function of the cathepsin protein through the glycosyltransferase 6 domain containing 1 (*GLT6D1*) protein [8].

Diseases caused by mutations in the *EDA1* gene show XLD (X-linked dominant) or XLR (X-linked recessive) inheritance and can lead to the development of two different clinical entities: one with only dental symptoms, this is tooth agenesis 1 (STHAGX1; OMIM 303500), the other entity is ectodermal dysplasia 1, also known as Christ-Siemens-Touraine syndrome (ECTD1; OMIM 305100), the characteristic triad of which is reduced sweating, reduced or missing fur and dental symptoms [9]. We performed the genetic examination of a Hungarian patient who had the complete triad of ECTD1 [9]. The identified variant affects the tumor necrosis factor (TNF α) binding domain of the *EDA1* protein; presumably through the dysfunction of this domain it can contribute to the induction of pathological cell biological processes, which cause the disease [9]. The interesting feature about the missense mutation identified on the *EDA1* gene (p.Val324Glu) is that heterozygous carrier women are almost completely symptom-free: their coat and hair are normal and sweating is in the normal range (Table 6). Only the presence of cone-shaped teeth can draw attention to the heterozygous carrier status [9].

Table 6. Clinical symptoms and genetic screening results of the Hungarian family affected by ECTD1 [9].

Patient ID No.	1.	2.	3.
Phenotype	ECTD1	ECTD1	ECTD1
Hypotrichosis	+	-	-
Hypohidrosis	+	-	-
Missing teeth	+	-	-
Conical shaped teeth	+	+	+
p.Val324Glu variant of the <i>EDA1</i>	hemizygote	heterozygote	heterozygote

Summary

As a result of the explosive technological development in the field of genetics and genomics over the past decades, enormous progress has been made in understanding the genetic background of rare monogenic dental diseases with cutaneous manifestations. Reviewing the clinical and genetic characteristics of these diseases may raise the attention of the clinicians and helps the initiation of genetic screening. A study investigating the monogenic skin diseases (genodermatoses) with teeth abnormalities have already been published [10]. However, our study is unique in the aspects that we have focused primarily on monogenic dental diseases and we have performed a comprehensive review of the OMIM data and incorporated cases from our own clinical practice.

The reviewed examples of the *WNT10A*, *CTSC* and *EDA1* phenotypic spectra clearly illustrate that the variants of the same disease-associated gene can

lead to the development of variable clinical entities. If the possibility of a rare, genetically determined dental-cutaneous disease arises on the basis of the above summarized dental and cutaneous manifestations, then it is definitely worth referring the patient to genetic counseling and possibly genetic testing.

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Additional information

Conflict of interest

The authors have declared that no competing interests exist.

Ethical statements

Written informed consent was obtained from the HMS patient (Fig. 6) to publish the picture of her hands.

The authors declared that no clinical trials were used in the present study.

The authors declared that no experiments on humans or human tissues were performed for the present study.

The authors declared that no informed consent was obtained from the humans, donors or donors' representatives participating in the study.

The authors declared that no experiments on animals were performed for the present study.

The authors declared that no commercially available immortalised human and animal cell lines were used in the present study.

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Author contributions

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Data availability

All of the data that support the findings of this study are available in the main text.

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