

Ectodermal Dysplasia (generic term)

Authors: Doctor Kathleen Mortier¹, Professor Georges Wackens¹

Creation date: September 2004

Scientific Editor: Professor Antonella Tosti

¹Department of stomatology and maxillofacial surgery, AZ VUB Brussels, Belgium
kathleen.mortier@tiscali.be

Definition

Clinical classification of Ectodermal dysplasias

References

Definition

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures, such as hypohidrosis, hypotrichosis, onychodysplasia and hypodontia or anodontia.

About 160 clinically and genetically distinct hereditary ectodermal dysplasias have been cataloged. In the early seventies there existed no definition and no classification. Freire-Maia and Pinheiro tried to put some order in the field of ectodermal dysplasias. Firstly, the group should be defined before an attempt was made to list its conditions. Secondly, the group was so large that it was necessary to split it into several subgroups. So they decided that an ED should present any two of the signs that affected the four structures widely mentioned by the authors who studied the classic EDs – hair, teeth, nails and sweat glands – with or without any other sign (see blow). The system is arbitrary without biological relevance to the pathogenesis and genetics of the specific disorder. However, classification based on clinical signs and symptoms is all that has been available until recently, since the pathogenesis and molecular genetics of the disorder are largely unknown.

Clinical classification of Ectodermal dysplasias

(Pinheiro and Freire-Maia, 1994)

Conditions	Unknown cause						
	AD	AR	XL	?	AD?	AR?	XL?
Subgroup 1-2-3-4							
1. Christ-Siemens-Touraine (CST) syndrome (MIM 305100; BDE 0333; POS 3208; FMP 1)			XR				
2. AR hypohydrotic ED (MIM 224900; BDE 3120; FMP 2)		X					
3. Focal dermal hypoplasia (FDH) (MIM 305600; BDE 0281; POS 3204; FMP 3)			XD ^a				
4. Xeroderma-talipes-enamel defect (XTE) (POS 4054; FMP 4)		X					
5. Rosselli-Gulienetti syndrome (MIM 225000; BDE 0179; POS 3141; FMP 5) ^b		X					
6. Dyskeratosis congenita, Scoggins type (MIM 127550; BDE 2024; POS 3191; FMP 6)	X						
7. Dyskeratosis congenita, AR (MIM 224230; BDE 2024; POS 3191; FMP 6)		X					

Conditions	AD	AR	XL	?	AD?	AR?	XL?
8. Dyskeratosis congenita, Zinsser-Cole-Engman type (MIM 305000; BDE 2024; POS 3191; FMP 6)			XR				
9. Pachyonychia congenita, Jadassohn-Lewandowsky type (MIM 167200; BDE 0789; POS 4181; FMP 7)	X						
10. Pachyonychia congenita, Jackson-Lawler type (MIM 167210; BDE 2905)	X						
11. Rapp-Hodgkin syndrome (MIM 129400; BDE 3056; POS 3137; FMP 8)	X						
12. Ectrodactyly-ED-cleft lip/palate (EEC) syndrome (MIM 129900; BDE 0337; POS 3211; FMP9)	X						
13. Ankyloblepharon-ectodermal defect-cleft lip/palate (AEC) syndrome (MIM 106260; BDE 2590; POS 3524; FMP 10)	X						
14. Zanier-Roubicek syndrome (MIM 129490; FMP11)	X						
15. Tricho-onycho-dental (TOD) dysplasia (POS 4730; FMP 12)	X						
16. ED with hypotrichosis, hypohidrosis, defective teeth, and unusual dermatoglyphics (MIM 129200; BDE 0102; POS 3822; FMP 13) ^c					X		XD
17. Carey syndrome (FMP 14)				X			
18. Camarena syndrome (FMP 15)					X		XD
19. Keratitis-ichthyosis-deafness (KID) (MIM 242150; BDE 2861; POS 3528; FMP16)		X					
20. Anonychia with flexural pigmentation (MIM 106750; FMP 17)					X		XD
21. Hypohidrotic ED with papillomas and acanthosis nigricans (FMP 18)						X	
22. Odonto-onycho-hypohidrotic dysplasia with midline scalp defect (MIM 129550; BDE 2833; POS 4041; FMP 19)	X						
23. Odonto-onycho-dermal dysplasia (MIM 257980; BDE 2618; POS 3609; FMP 20)		X					
24. Tricho-odonto-onycho-hypohidrotic dysplasia with cataract (FMP 21) ^d						X	
25. Papillon-Lefèvre syndrome (MIM 245000; BDE 0494; POS 3647; FMP 22; Kotzot and Pfeiffer [1993])		X					
26. Hypomelanosis of Ito, AD (MIM 146150; BDE 2264; POS 3525; FMP 23)	X						
27. Hypomelanosis of Ito, XD (MIM 146150; BDE 2264; POS 3525; Koiffmann et al. [1993])			XD				
28. ED with cardiac and skeletal abnormalities [Lipson, 1988]				X			
29. Zlotogora-Ogur syndrome (MIM 225000; BDE 3126; POS 3625)		X					
30. Naegeli-Franceschetti-Jadassohn syndrome (MIM 161000; BDE 0703; POS 4118; FMP 1 in subgroup 2-4)	X						
Total of subgroup 1-2-3-4	11	8	4	2	3	2	3
Subgroup 1-2-3							
1. Rothmund-Thomson syndrome (MIM 268400; BDE 2037; POS 3383; FMP1)		X					
2. Fischer-Jacobsen-Clouston syndrome (MIM 129500; BDE 0334; POS 3209; FMP 2)	X						
3. Coffin-Siris syndrome (MIM 135900; BDE 2025; POS 3151; FMP 3) ^e					X	X	XD
4. Odontotrichomelic syndrome (MIM 273400; BDE 2887; POS 3355; FMP 4)		X					
5. Trichodontoosseous (TDO) syndrome (MIM 190320; BDE 0965 POS 3414; FMP 5,6,7) ^f	X						
6. Incontinentia pigmenti (MIM 308300; BDE 0526; POS 3265; FMP 8)			XD ^a				
7. Cranioectodermal dysplasia (MIM 218330; BDE 2127; POS 3409; FMP 9; Lammer et al. [1993])						X	
8. Fried's tooth and nail syndrome (FMP 10)		X					

Conditions	AD	AR	XL	?	AD?	AR?	XL?
9. Hypodontia-nail dysgenesis (MIM 189500; BDE 0511; POS 3261; FMP 11)	X						
10. Dentooculocutaneous syndrome (MIM 200970; BDE 2093; POS 4381; FMP 12)		X ^g					
11. Trichorhinophalangeal (TRP) syndrome, Murdoch-Gorlin type (MIM 190350; BDE 0966; POS 3415; FMP 13)	X						
12. Trichorhinophalangeal (TRP) syndrome, Giedion type (MIM 275500; BDE 0966; POS 3415; FMP 13)		X					
13. Ellis-van Creveld syndrome (MIM 225500; BDE 0156; POS 3066; FMP 14)		X					
14. Cystic eyelids-palmoplantar keratosis-hypodontia-hypotrichosis (POS 4889; FMP 15; Burket et al. [1984]; Font et al. [1986] ^h)		X					
15. Šalamon-Miličević syndrome (FMP 16)		X					
16. Tricho-oculo-dermo-vertebral syndrome (POS 4955; FMP 17; Stratton et al. [1993])		X					
17. Oculodentodigital (ODD) syndrome II (FMP 18)				X			
18. Arthrogyposis and ED (POS 3960; FMP 19)				X			
19. Tricho-odonto-onycho-dermal syndrome (FMP 20)				X			
20. Tricho-odonto-onychia dysplasia (MIM 275450; BDE 2889; POS 3603; FMP 21)						X	
21. Odonto-onychodysplasia-alopecia (BDE 2890; POS 3422; FMP 22)		X					
22. Schinzel-Giedion syndrome (MIM 269150; BDE 2123; POS 3025; FMP 23)		X					
23. Growth retardation-alopecia-pseudoanodontia-optic atrophy (GAPO) (MIM 230740; BDE 2293; POS 3640; FMP 24; Sayli and Gül [1993])		X					
24. ED with pillous anomaly and syndactyly (FMP 25)		X					
25. Osteosclerosis and ED (POS 4382; FMP 26; Blau [1987])		X					
26. Dermoodontodysplasia (MIM 125640; BDE 2763; FMP 27)	X						
27. Tricho-odonto-onychodysplasia with pili torti (FMP 28)					X		XD
28. Mesomelic dwarfism-skeletal abnormalities-ED (FMP 29; Brunoni et al. [1984])				X			
29. ED syndrome with tetramelic deficiencies (FMP 30)				X			
30. Pilo-dento-ungular dysplasia with microcephaly (BDE 2636; POS 4391)		X					
31. Tricho-dermodyplasia-dental defects (BDE 2903; POS 4226)				X			XD
32. Oculotrichodysplasia (OTD) (MIM 257960)		X					
33. Brittle hair, intellectual impairment, decreased fertility, and short stature (BIDS) syndrome (MIM 234050; BDE 2559; POS 3456) ⁱ		X					
34. Dolichocephaly, dental defects, trichodysplasia (F. Carnevale and M.G. Greco, 1983, personal communication)	X						
35. Hypoplasia of nails, malformations of hands and feet, curly hair, microdontia, seizures (POS 4619; Cortes and Lacassie [1986])				X			
36. Sparse hair-short stature-hypoplastic thumbs-hypodontia-skin anomaly [Winter et al., 1988]					X		XD
37. Hypotrichosis with pili bifurcati [Beemer et al., 1988] ^j						X	
38. ED-facial alterations-polydactyly (MIM 129540; POS 4542)				X			
39. ED with amastia and athelia (MIM 129510)	X						
40. Cardio-facio-cutaneous (CFC) syndrome (MIM 115150; BDE 2587; POS 3627; Lopez-Rangel et al. [1993])						X	
41. ADULT syndrome (POS 5233; Propping and Zerres [1993])	X						
42. Alopecia-contractures-dwarfism (ACD)-mental retardation (MIM 203550; BDE 2782; POS 3562; FMP 8 in subgroup 1-3)		X					

Conditions	AD	AR	XL	?	AD?	AR?	XL?
43. ED, Margarita type (MIM 225060; POS 4936)		X					
Total of subgroup 1-2-3	8	19	1	7	4	5	4
Subgroup 1-2-4							
1. Regional ED with total bilateral cleft (FMP 1)				X			
2. Leucomelanoderma-infantilism-mental retardation-hypodontia-hypotrichosis (MIM 246500; BDE 0105; POS 3827; FMP 2)		X					
3. Premolar aplasia-hyperhidrosis-canities prematura (PHC) (MIM 112300; BDE 0493; POS 3041; FMP 3)	X						
4. Lenz-Passarge dysplasia (FMP 5)			XD				
5. Wesser-Vistnes ED with palatal paralysis (FMP 6)				X			
6. Alopecia-anosmia-deafness-hypogonadism (AADH) (MIM 147770; BDE 2765; POS 3595)	X						
7. Hypohidrotic ED with focal sweating [Gorlin, 1988, pp 128-129]						X	XR
8. Cleft lip/palate, ED, acral anomalies (POS 3783; Richieri-Costa et al. [1992])		X					
Total of subgroup 1-2-4	2	2	1	2	0	1	1
Subgroup 1-3-4							
1. Fischer-Volavsek syndrome (FMP 1)	X						
2. Trichodysplasia-onychogryposis-hypohidrosis-cataract (FMP 2)				X			
3. Alopecia-onychodysplasia-hypohidrosis-deafness (FMP 3)				X			
4. Alopecia-nail dystrophy-ophthalmic complication-thyroid dysfunction-hypohidrosis-ephelides-enteropathy-respiratory tract infections (ANOTHER) (MIM 225050; FMP 6)						X	
5. ED with severe mental retardation (FMP 7)				X			
6. Dermotrichic syndrome (FMP 9)			XR	X			
7. Alopecia-skin atrophy-anonychia-tongue defects (BDE 2842; POS 4104)				X			
8. ED with digital and eye anomalies [Viljoen and Winship, 1988]				X			
Total of subgroup 1-3-4	1	0	1	5	0	1	0
Subgroup 2-3-4							
1. Amelo-onycho-hypohidrotic dysplasia (MIM 104570; BDE 0045; POS 3022; FMP 1)	X						
Total of subgroup 2-3-4	1	0	0	0	0	0	0
Subgroup 1-2							
1. Orofaciodigital (OFD) syndrome, type I (MIM 311200; BDE 0770; POS 3347; FMP 1)			XD ^a				
2. Oculodentodigital (ODD) syndrome (MIM 164200; BDE 0737; POS 3341; FMP 2)	X						
3. Oculodentoosseous dysplasia (ODOD) (MIM 257850)		X					
4. Hallermann-Streiff syndrome (MIM 234100; BDE 0738; POS 3342; FMP 3) ^e					X	X	XD
5. Gorlin-Chaudry-Moss syndrome (MIM 233500; BDE 0440; POS 3173; FMP 4)						X	
6. ED with neurosensory deafness (MIM 224800; POS 4489; FMP 5)		X					
7. Gingival fibromatosis-sparse hair-malposition of teeth (FMP 6)		X					
8. Hypertrichosis and dental defects (MIM 145700; BDE 0507; FMP 7) ^k	X						
9. Gingival fibromatosis-hypertrichosis (MIM 135400; BDE 0410; POS 3323; FMP 8; Bondeson and Miles [1993])	X						

Conditions	AD	AR	XL	?	AD?	AR?	XL?
10. Twisted hairs and enamel hypoplasia (MIM 261900; POS 3876; FMP 9)	X						
11. Walbaum-Dehaene-Schlemmer syndrome (FMP 10)		X					
12. Brachymetapody-anodontia-hypotrichosis-albinoidism (MIM 211370; BDE 2078; POS 3044; FMP 11)						X	
13. Johanson-Blizzard syndrome (MIM 243800; BDE 2026; POS 3269; FMP 13)		X					
14. Trichodental dysplasia (FMP 14; Kersey, 1987)	X						
15. Ectrodactyly with ED without cleft lip/palate (MIM 129810)	X						
16. ED-ectrodactyly-macular dystrophy (EEM) (MIM 225280; BDE 2793; POS 3580)		X					
17. Cleft lip/palate-oligodontia-syndactyly-hair defects (BDE 2898; POS 4220)					X		XD
18. Zurich neuroectodermal syndrome (MIM 280000; POS 3605)						X	
19. Pilodental dysplasia with refractive errors (MIM 262020; BDE 2813; POS 4322)						X	
20. Uncombable hair-retinal pigmentary dystrophy-juvenile cataract-brachymetacarpia [Bork et al., 1987]	X						
21. Clefing-ectropion-dental anomaly (MIM 119580; BDE 2759; POS 3667)	X						
22. Ichthyosis, brittle hair, impaired intelligence, decreased fertility, short stature (IBIDS) (MIM 242170; BDE 2559; POS 3634)		X					
23. Dubowitz syndrome (MIM 223370; BDE 0299; POS 3187)		X					
24. Growth retardation, ocular abnormalities, microcephaly, brachydactyly, oligophrenia (GOMBO) syndrome (MIM 233270; POS 4939)						X	
25. Cartilage-hair hypoplasia (CHH) (MIM 250250; BDE 0653; POS 3061)		X					
26. Cataract, hypertrichosis, mental retardation (CAHMR) syndrome (MIM 211770)		X					
27. Trichodysplasia and amelogenesis imperfecta (Angelos and Jorgenson, 1993)					X		XD
28. Oligodontia, taurodontia, and sparse hair growth (MIM 272980; POS 3790 and 4029)						X	
Total of subgroup 1-2	8	10	1	0	3	7	3
Subgroup 1-3							
1. Hairy elbows (MIM 139600; FMP 1)		X					
2. Palmoplantar hyperkeratosis and alopecia (MIM 104100; FMP 2)	X						
3. Curly hair-ankyloblepharon-nail dysplasia syndrome (CHANDS) (MIM 214350; BDE 3039; POS 3845; FMP 3)		X					
4. Onychotrichodysplasia and neutropenia (MIM 258360; BDE 2331; POS 3490; FMP 4)		X					
5. Pili torti and onychodysplasia (POS 4225; FMP 5)	X						
6. Tricho-onychodysplasia-xeroderma (BDE 2892; FMP 7)		X					
7. Sabinas brittle hair and mental deficiency syndrome (MIM 211390; BDE 2559; POS 4423; FMP 9)		X					
8. Syndrome of accelerated skeletal maturation, failure to thrive and peculiar face (FMP 10)						X	XR
9. Lymphedema-hypoparathyroidism syndrome (MIM 247410; BDE 2801; POS 3599; FMP 11) ^e						X	XR
10. Bartsocas-Papas syndrome (MIM 263650; BDE 3233)		X					
11. Digitorenalcerebral (DRC) syndrome (MIM 222760; BDE 2792; POS 3446)		X					
12. Onychodystrophy and aplasia/hypoplasia of distal phalanges [Cooks et al., 1985]	X						

Conditions	AD	AR	XL	?	AD?	AR?	XL?
13. Trichomegaly-mental retardation-dwarfism-retinal pigmentary degeneration (MIM 275400; BDE 2294; POS 3442)						X	
14. Ichthyosis-alopecia-eclabion-ectropion-mental retardation (MIM 242510; POS 4300)						X	
15. ED with skin anomalies and mental retardation [Halal et al., 1991]		X					
16. Alveolar synechia-ankyloblepharon-ectodermal disorders [Ohishi et al., 1991] ^l		X					
17. Polyposis-skin pigmentation-alopecia-nail defects (MIM 175500; BDE 3040; POS 3844)				X			
18. Pollitt syndrome (MIM 275550) ^m						X	
19. Neuroichthyosis-hypogonadism (MIM 308200; BDE 0741; POS 3850; FMP 5 in subgroup 1-3-4)						X	XR
Total of subgroup 1-3	3	9	0	1	0	6	3
Subgroup 1-4							
1. Focal facial dermal dysplasia (MIM 136500; FMP 1)	X						
2. Facial ED (MIM 227260; BDE 2095; POS 3407; FMP 1)		X					
3. Tricho-facio-hypohidrotic syndrome [Antley et al., 1976]						X	XR
4. Dry skin and extraneous arelae [Freire-Maia and Chautard-Freire-Maia, 1990]	X						
5. Short stature-kidney insufficiency-ophthalmological anomaly-growth retardation-ED (SKORED) [Greenstein et al., 1985]						X	XR
Total of subgroup 1-4	2	1	0	0	0	2	2
Subgroup 2-3							
1. Deafness-onychoosteodystrophy-mental retardation (DOOR) (MIM 220500; BDE 0262; POS 3733; FMP 1)		X					
2. Deafness and onychodystrophy (MIM 124480; BDE 2034; POS 3581; FMP 3)	X						
3. Ectodermal defect with skeletal abnormalities (FMP 2)				X			
4. Odontoonychodysplasia (POS 4102; FMP 5) ⁿ	X						
5. Kirghizian dermato-osteolysis (MIM 221810; BDE 3044; POS 3604; FMP 6)						X	
6. Corneodermatoosseous (CDO) syndrome (MIM 122440; BDE 2760; POS 3753)	X						
7. Lacrimo-auriculo-dento-digital (LADD) syndrome (MIM 149730; BDE 2180; POS 3546)	X						
8. Hearing loss, sensorineural, with enamel hypoplasia and nail defects (MIM 234580)						X	
Total of subgroup 2-3	4	1	0	1	0	2	0
Subgroup 2-4							
1. Amelo-cerebro-hypohidrotic syndrome (MIM 226750; BDE 0044; POS 3021; FMP 3; Zlotogora et al. [1993])		X					
2. Hypohidrotic ED with mydriasis, iris atrophy, and mental retardation [Beyer et al., 1979]					X		
Total of subgroup 2-4	0	1	0	0	1	0	0
Subgroup 3-4							
1. Absence of dermal ridge patterns, onychodystrophy and palmoplantar anhidrosis (FMP 1) ^o	X						
2. Pachyonychia congenita, AR type (MIM 260130)		X					
Total of subgroup 3-4	1	1	0	0	0	0	0

AD: Autosomal dominant inheritance

AR: Autosomal recessive inheritance

XL: X-linked inheritance

The number after MIM is the number of the respective condition in the [McKusick catalog](#) [1992]; BDE refers to the number in the Birth Defects Encyclopedia [Buyse, 1990]; POS is the [POSSUM number](#); FMP followed by a number refers to the number of the condition in the respective subgroup in our book [Freire-Maia and Pinheiro, 1984].

^a With lethality in males.

^b According to MIM 225000, it includes Zlotogora-Ogur syndrome (subgroup 1-2-3-4, no. 29).

^c The family described by Basan [1965] possibly has another condition.

^d MIM 268400 as Rothmund-Thomson syndrome.

^e Etiological heterogeneity?

^f Three clinical types (possibly different AD entities) are generally referred to: Lichtenstein type (TDO I), Leisti-Sjöblom type (TDO II), and Shapiro type (TDO III).

^g With possible mild manifestation in heterozygotes.

^h MIM 245000 as Papillon-Lefèvre syndrome.

ⁱ BIDS may include Sabinas brittle hair syndrome (subgroup 1-3, no. 7), Pollitt syndrome (subgroup 1-3, no. 18) and IBIDS (subgroup 1-2, no. 23).

^j No information on the sex of the two affected children is provided.

^k Heterogeneity is possible. Most of the cases are described as not having teeth defects.

^l MIM 119500 as popliteal pterigium syndrome.

^m "The enamel of the teeth, though intact, was thin" (Pollitt et al., 1968).

ⁿ MIM 167200 as pachyonychia congenita, Jadassohn-Lewandowsky type.

^o MIM 129200 as Basan syndrome.

References

Pinheiro M., Freire-Maia N.: Ectodermal dysplasias: a clinical classification and a causal review. *Am. J. Med Genet.* (1994) 53: 153-162

Freire-Maia N, Lisboa-Costa T, Pagnan NA. R Ectodermal dysplasias: how many? *Am J Med Genet.* 2001;104 :84.

Pinheiro M, Freire-Maia N. [Ectodermal dysplasias--a general view] *Rev Assoc Med Bras.* 1992 Oct-Dec;38(4):221-4.

Freire-Maia N, Pinheiro M. Ectodermal dysplasias--some recollections and a classification. *Birth Defects Orig Artic Ser.* 1988;24:3-14.