

| CODE  | PATHOLOGIE                                                | Omim   | Individus | Familles | Atteints |
|-------|-----------------------------------------------------------|--------|-----------|----------|----------|
| AEZ   | ACRODERMATITIS ENTEROPATHICA                              | 201100 | 4         | 1        | 3        |
| AGA   | ACROGERIA                                                 | 201200 | 2         | 1        | 1        |
| ALF   | ALOPECIA                                                  | 104110 | 3         | 1        | 3        |
| AN    | ACANTHOSIS NIGRICANS                                      | 100600 | 10        | 1        | 4        |
| ARF   | ICHTHYOSIS CONGENITA, HARLEQUIN FETUS TYPE                | 242500 | 12        | 3        | 3        |
| BIE   | <b>BULLOUS CONGENITAL ICHTHYOSIFORM ERYTHRODERMA</b>      | 113800 | 5         | 1        | 1        |
| BLM   | BLOOM SYNDROME                                            | 210900 | 3         | 1        | 1        |
| BZX   | BAZEX-DUPRE-CHRISTOL SYNDROME                             | 301845 | 4         | 1        | 3        |
| CBB   | COLLODION BABY                                            | 242300 | 765       | 129      | 167      |
| CDS   | CHANARIN-DORFMAN SYNDROME                                 | 275630 | 52        | 15       | 18       |
| CHBL  | CHILBLAIN LUPUS 1                                         | 610448 | 2         | 1        | 2        |
| CHS1  | CHEDIAK HIGASHI SYNDROM                                   | 214500 | 7         | 2        | 3        |
| CMS   | ICHTHYOSIS HYSTRIX, CURTH-MACKLIN TYPE                    | 146590 | 22        | 3        | 9        |
| CPP   | CALLOSITIES HEREDITARY PAINFUL                            | 114140 | 2         | 1        | 2        |
| CS    | COCKAYNE SYNDROME                                         | 216400 | 14        | 8        | 7        |
| CTLX  | CUTIS-LAXA                                                | 123700 | 115       | 32       | 45       |
| CZC   | CROUZON SYNDROME                                          | 123500 | 1         | 1        | 1        |
| DAE   | ECTODERMAL DYSPLASIA ANHIDROTIC                           | 224900 | 30        | 9        | 13       |
| EBCT  | EPIDERMOLYSIS BULLOSA, COCKAYNE-TOURAIN TYPE              | 131800 | 11        | 4        | 5        |
| EBDHS | EPIDERMOLYSIS BULLOSA DYSTROPHICA, HALLOPEAU-SIEMENS TYPE | 226600 | 256       | 65       | 79       |
| EBDI  | EPIDERMOLYSIS BULLOSA INVERSA DYSTROPHICA                 | 226450 | 6         | 1        | 1        |
| EBDM  | EPIDERMOLYSIS BULLOSA HERPETIFORMIS, DOWLING-MEARA TYPE   | 131760 | 104       | 21       | 40       |
| EBDP  | EPIDERMOLYSIS BULLOSA DYSTROPHICA, PASINI TYPE            | 131750 | 47        | 11       | 18       |
| EBDR  | EPIDERMOLYSIS BULLOSA DYSTROPHICA (MINOR FORM)            |        | 11        | 4        | 4        |
| EBJ   | EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN        | 226650 | 144       | 20       | 12       |
| EBPR  | EPIDERMOLYSIS BULLOSA PRETIBIAL                           | 131850 | 5         | 1        | 3        |

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| EBS   | EPIDERMOLYSIS BULLOSA SIMPLEX                                 | 601001 | 20   | 4   | 8   |
| EBSK  | EPIDERMOLYSIS BULLOSA SIMPLEX, KOEBNER TYPE                   | 131900 | 14   | 3   | 5   |
| EBWC  | EPIDERMOLYSIS BULLOSA SIMPLEX, WEBER-COCKAYNE TYPE            | 131800 | 19   | 5   | 11  |
| ECIB  | ICHTHYOSIFORME ERYTHRODERMA CONGENITAL                        | 242100 | 80   | 25  | 32  |
| ECIS  | ICHTHYOSIFORM ERYTHRODERMA CONGENITAL NONBULLOUS              | 242100 | 30   | 7   | 8   |
| ED4   | EHLERS-DANLOS SYNDROME TYPE IV                                | 130050 | 17   | 4   | 9   |
| EDC   | ECTODERMAL DYSPLASIA HIDROTIC, CLOUSTON SYNDROME              | 129500 | 66   | 7   | 28  |
| EDS1  | EHLERS-DANLOS SYNDROME                                        | 130000 | 255  | 155 | 191 |
| EKV   | ERYTHROKERATODERMIA VARIABILIS                                | 133200 | 11   | 2   | 2   |
| EV    | EPIDERMODYSPLASIA VERRUCIFORME                                | 226400 | 226  | 32  | 40  |
| FDH   | FOCAL DERMAL HYPOPLASIA-GOLTZ SYNDROME                        | 305600 | 8    | 3   | 4   |
| GO    | GERODERMA OSTEODYSPLASTICA                                    | 231070 | 5    | 1   | 1   |
| GS    | <b>GITELMAN SYNDROME</b>                                      | 263800 | 1    | 1   | 1   |
| HGPS  | HUTCHINSON-GILFORD PROGERIA SYNDROME                          | 176670 | 3    | 3   | 3   |
| HKP   | FLEXURES KERATODERMA                                          |        | 5    | 2   | 1   |
| IAD   | ICHTHYOSIS AUTOSOMAL DOMINANT                                 | 146750 | 15   | 3   | 11  |
| ICH   | ICHTHYOSIS CONGENITA                                          | 242300 | 1173 | 248 | 351 |
| ICHBS | ICHTHYOSIS BULLOUS EXFOLIATIVA                                | 146800 | 3    | 1   | 1   |
| ICHHP | ICHTHYOSIS AND PAPILLOMATOSIS                                 |        | 2    | 1   | 0   |
| IFAP  | ICHTHYOSIS FOLLICULARIS,ATRICHIA,AND PHOTOPHOBIA SYNDROME     | 308205 | 13   | 2   | 3   |
| IV    | ICHTHYOSIS VULGARIS                                           | 146700 | 8    | 2   | 2   |
| KAP   | KAPOSI SARCOMA, MULTIPLE IDIOPATHIC PIGMENTED HEMANGIOSARCOMA | 148000 | 2    | 1   | 2   |
| KDAC  | ACANTHOLYTIC KERATOSIS AND PALMOPLANTARIS KERATODERMA         |        | 4    | 1   | 1   |
| KEC   | KERATOLYSIS EXFOLIATIVA CONGENITA                             | 270300 | 9    | 2   | 3   |
| KFSD  | KERATOSIS FOLLICULARIS SPINULOSA DECALVANS                    | 308800 | 7    | 1   | 2   |
| KID   | KERATITIS/ICHTHYOSIOS/DEAFNESS SYNDROME                       | 148210 | 9    | 2   | 2   |
| KLS   | KLICK SYNDROME/KERATOSIS LINEARIS WITH ICHTHYOSIS CONGENITA   | 601952 | 9    | 1   | 1   |
| KPP   | UNDEFINED PALMO-PLANT KERATODERMA                             |        | 50   | 9   | 21  |
| KPPBF | KERATOSIS PALMOPLANTARIS PALPULOSA BUSCKE-FISCHER-BRAUER TYPE | 148600 | 4    | 2   | 3   |
| KPPBU | KPP DE BURNAUHER-FUEHS                                        |        | 1    | 1   | 1   |
| KPPF  | AXIONAL NEUROPATHY WITH PALMOPLANTAIR KERATODERMA             | 148360 | 2    | 1   | 1   |
| KPPJL | PACHYONYCHIA CONGENITA                                        | 167210 | 6    | 1   | 4   |
| KPPM  | MELEDA DISEASE                                                | 248300 | 310  | 49  | 95  |
| KPPPL | PAPILLON LEFEVRE SYNDROME                                     | 245000 | 58   | 14  | 24  |

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| KPPTU | UNNA-THOST DESAESE, NON EPIDERMOLYTIC                          | 600962 | 12   | 1   | 7   |
| KPPV  | PALMOPLANTAIR KERATOSIS VORNER TYPE (UNNA-THOST EPIDERMOLYTIC) | 144200 | 35   | 1   | 16  |
| KS    | KINDLER SYNDROME, POIKIODERMA                                  | 173650 | 29   | 9   | 12  |
| LIP   | LENTIGINOSIS, INHERITED PATTERNED                              | 151001 | 1    | 1   | 1   |
| LIP0  | LIPOMATOSIS MULTIPLE                                           | 151900 | 2    | 2   | 2   |
| MCL   | LEIOMYOMA, MULTIPLE CUTANEUS                                   | 150800 | 8    | 1   | 4   |
| NBC   | BASAL CELL NAEVUS SYNDROME                                     | 109400 | 14   | 3   | 7   |
| NELI  | NON ERYTHRODERMIC LAMELLAR ICHTHYOSIS                          | 604777 | 2    | 1   | 1   |
| NET   | NETHERTON SYNDROME                                             | 256500 | 36   | 9   | 10  |
| NYS1  | NYSTAGMUS CONGENITAL                                           | 310700 | 3    | 1   | 2   |
| PC1   | PACHYONYCHIA CONGENITA, TYPE 1                                 | 167200 | 5    | 1   | 1   |
| PCT   | PORPHYRIA CUTANEA TARDA                                        | 176100 | 2    | 2   | 2   |
| PPPD  | PLANTARIS PALMARIS DISSEMINATA                                 | 175850 | 4    | 2   | 3   |
| PRP   | PITYRIASIS RUBIA PILASIS                                       | 173200 | 5    | 1   | 2   |
| PSEK  | ERYTHROKERATODERMIA PROGRESSIVE SYMETRIC                       | 602036 | 40   | 4   | 14  |
| PSO   | PSORIASIS                                                      | 177900 | 2078 | 202 | 822 |
| PSS   | FAMILIAL CONTINUOUS SKIN PEELING                               | 270300 | 18   | 5   | 6   |
| PXE   | PSEUDOXANTHOMA ELASTICUM                                       | 177850 | 20   | 5   | 4   |
| RPAF  | RETICULAR PIGMENTED ANOMALY OF FLEXURES                        | 179850 | 11   | 2   | 7   |
| RTS   | ROTHMUND-THOMSON SYNDROME                                      | 268400 | 14   | 5   | 7   |
| SLS   | SJOGREN-LARSSON SYNDROME                                       | 270200 | 7    | 3   | 3   |
| SM    | STEATOCYSTOMA MULTIPLEX                                        | 184500 | 61   | 13  | 35  |
| SY    | SYRINGOMAS MULTIPLE                                            | 186600 | 8    | 3   | 4   |
| TCHE  | TRICHOEPITHELIOMAS MULTIPLE DESMOPLASTIC                       | 190345 | 14   | 2   | 5   |
| TTD   | TRICHOTHIODYSTROPHY                                            | 601675 | 45   | 10  | 12  |
| VOH   | VOHWINKEL SYNDROME                                             | 604117 | 20   | 3   | 4   |
| VTG   | VITILIGO                                                       | 193200 | 26   | 4   | 9   |
| WOH   | WOOLLY HAIR                                                    | 194300 | 6    | 1   | 4   |
| WRN   | SYNDROMES DE WERNER                                            | 277700 | 6    | 1   | 2   |
| XP    | XERODERMA PIGMENTOSUM                                          | 278730 | 65   | 15  | 23  |
| XPA   | XERADERMA PIGMENTOSUM GROUPE A                                 | 278700 | 4    | 1   | 1   |
| XRI   | ICHTHYOSIS X LINKED                                            | 308100 | 62   | 13  | 22  |