Panel de Dermatología

Es nuestra solución para pacientes que presentan trastornos de la piel. Nuestro panel incluye genes para hipotricosis, epidermólisis ampollosa e ictiosis congénita, entre otros.

*Para melanoma, hay que mirar el panel de Cáncer o el panel Cáncer Integral

Nº de genes:	152
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

Síndromes y trastornos comunes cubiertos

- Ictiosis congénita
- Cutis laxo
- Epidermólisis ampollosa
- Ictiosis extendida
- Hipotricosis no sindrómica

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ABCA12	607800	Ichthyosis, congenital, AR 4A;Ichthyosis, congenital, AR 4B (harlequin)	AR
ABHD5	604780	Chanarin-Dorfman syndrome	AR
ACD	609377	Dyskeratosis congenita, AD 6;?Dyskeratosis congenita, AR 7	AD, AR
ALAD	125270	Porphyria, acute hepatic;Lead poisoning, susceptibility to	AR

ALAS2	301300	Protoporphyria, erythropoietic, XL;Anemia, sideroblastic, 1	XL, XLR
ALDH18A1	138250	Cutis laxa, AD 3;Cutis laxa, AR, type IIIA;Spastic paraplegia 9B, AR;Spastic paraplegia 9A, AD	AD, AR
ALDH3A2	609523	Sjogren-Larsson syndrome	AR
ALOX12B	603741	Ichthyosis, congenital, AR 2	AR
ALOXE3	607206	Ichthyosis, congenital, AR 3	AR
AP1S1	603531	MEDNIK syndrome	AR
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
AP3D1	607246	Hermansky-Pudlak syndrome 10	AR
APCDD1	607479	Hypotrichosis 1	AD
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
ARSL	300180	Chondrodysplasia punctata, XLR	XLR
ATP6V0A2	611716	Cutis laxa, AR, type IIA;Wrinkly skin syndrome	AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	604310	Hermansky-pudlak syndrome 9	AR
CASP14	605848	Ichthyosis, congenital, AR 12	AR
CDSN	602593	Peeling skin syndrome 1;Hypotrichosis 2	AR, AD
CERS3	615276	Ichthyosis, congenital, AR 9	AR
CHST8	610190	Peeling skin syndrome 3	AR

CLDN1	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	AR
COL17A1	113811	Epithelial recurrent erosion dystrophy;Epidermolysis bullosa, junctional, localisata variant;Epidermolysis bullosa, junctional, non-Herlitz type	AD, AR
COL7A1	120120	Epidermolysis bullosa dystrophica, AR;Epidermolysis bullosa dystrophica, AD;Epidermolysis bullosa pruriginosa;Transient bullous of the newborn;Epidermolysis bullosa, pretibial;Toenail dystrophy, isolated;EBD, Bart type;EBD inversa	AR, AD, AD, AR
CPOX	612732	Harderoporphyria;Coproporphyria	AR, AD, AR
CSTA	184600	Peeling skin syndrome 4	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CYP4F22	611495	Ichthyosis, congenital, AR 5	AR
DKC1	300126	Dyskeratosis congenita, XL	XLR
DLL4	605185	Adams-Oliver syndrome 6	AD
DOCK6	614194	Adams-Oliver syndrome 2	AR
DSG1	125670	Keratosis palmoplantaris striata I, AD;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	AD, AR
DSG4	607892	Hypotrichosis 6	AR
DSP	125647	Cardiomyopathy, dilated, with woolly hair and keratoderma;Arrhythmogenic right ventricular dysplasia 8;Keratosis palmoplantaris striata II;Skin fragility-woolly hair syndrome;Epidermolysis bullosa, lethal acantholytic;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	AR, AD

DST	113810	Neuropathy, hereditary sensory and autonomic, type VI;Epidermolysis bullosa simplex, AR 2	AR
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
EBP	300205	MEND syndrome;Chondrodysplasia punctata, XLD	XLR, XLD
EDA	300451	Tooth agenesis, selective, XL 1;Ectodermal dysplasia 1, hypohidrotic, XL	XLD, XLR
EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, AR;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, AD;[Hair morphology 1, hair thickness]	AR, AD
EDARADD	606603	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, AD;Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, AR	AD, AR
EDN3	131242	Waardenburg syndrome, type 4B;Hirschsprung disease, susceptibility to, 4;Central hypoventilation syndrome, congenital	AD, AR, AD
EDNRB	131244	Waardenburg syndrome, type 4A;Hirschsprung disease, susceptibility to, 2;ABCD syndrome	AD, AR, AD, AR
EFEMP2	604633	Cutis laxa, AR, type IB	AR
ELN	130160	Cutis laxa, AD;Supravalvar aortic stenosis	AD
ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation;Stargardt disease 3;Spinocerebellar ataxia 34	AR, AD
EOGT	614789	Adams-Oliver syndrome 4	AR
EPG5	615068	Vici syndrome	AR
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2;Xeroderma pigmentosum, group D;Trichothiodystrophy 1, photosensitive	AR

ERCC3	133510	Xeroderma pigmentosum, group B;Trichothiodystrophy 2, photosensitive	AR
EXPH5	612878	Epidermolysis bullosa, nonspecific, AR	AR
FBLN5	604580	Cutis laxa, AR, type IA;Neuropathy, hereditary, with or without age-related macular degeneration;Macular degeneration, age-related, 3;?Cutis laxa, AD 2	AR, AD
FECH	612386	Protoporphyria, erythropoietic, 1	AR
FERMT1	607900	Kindler syndrome	AR
FLG	135940	Ichthyosis vulgaris;Dermatitis, atopic, susceptibility to, 2	AD, AR
FLG2	616284	Peeling skin syndrome 6	AR
GJB2	121011	Keratoderma, palmoplantar, with deafness;Keratitis-ichthyosis-deafness syndrome;Deafness, AD 3A;Hystrix-like ichthyosis with deafness;Bart-Pumphrey syndrome;Vohwinkel syndrome;Deafness, AR 1A	AD, AR, DD
GJB3	603324	Deafness, AD 2B;Erythrokeratodermia variabilis et progressiva 1;Deafness, digenic, GJB2/GJB3	AD, AD, AR, AR, DD
GJB4	605425	Erythrokeratodermia variabilis et progressiva 2	AD
GJB6	604418	Deafness, AR 1B;Deafness, digenic GJB2/GJB6;Ectodermal dysplasia 2, Clouston type;Deafness, AD 3B	AR, AR, DD, AD
GPR143	300808	Nystagmus 6, congenital, XL;Ocular albinism, type I, Nettleship-Falls type	XLR, XL
GTF2H5	608780	Trichothiodystrophy 3, photosensitive	
HMBS	609806	Porphyria, acute intermittent;Porphyria, acute intermittent, nonerythroid variant	AD

HPS1	604982	Hermansky-Pudlak syndrome 1	AR
HPS3	606118	Hermansky-Pudlak syndrome 3	AR
HPS4	606682	Hermansky-Pudlak syndrome 4	AR
HPS5	607521	Hermansky-Pudlak syndrome 5	AR
HPS6	607522	Hermansky-Pudlak syndrome 6	AR
HR	602302	Alopecia universalis;Atrichia with papular lesions;Hypotrichosis 4	AR, AD
ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	AR
ITGA6	147556	Epidermolysis bullosa, junctional, with pyloric stenosis	AR
ITGB4	147557	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa of hands and feet;Epidermolysis bullosa, junctional, with pyloric atresia	AR, AD
JUP	173325	Arrhythmogenic right ventricular dysplasia 12;Naxos disease	AD, AR
KCTD1	613420	Scalp-ear-nipple syndrome	AD
KDSR	136440	Erythrokeratodermia variabilis et progressiva 4	AR
KITLG	184745	[Skin/hair/eye pigmentation 7, blond/brown hair];Deafness, AD 69, unilateral or asymmetric;Hyperpigmentation with or without hypopigmentation	AD

KRT1	139350	Ichthyosis histrix, Curth-Macklin type;Epidermolytic hyperkeratosis;Keratosis palmoplantaris striata III;Ichthyosis, cyclic, with epidermolytic hyperkeratosis;Palmoplantar keratoderma, epidermolytic;Palmoplantar keratoderma, nonepidermolytic	AD, AD, AR
KRT10	148080	Ichthyosis, cyclic, with epidermolytic hyperkeratosis;Epidermolytic hyperkeratosis;Ichthyosis with confetti	AD, AD, AR
KRT14	148066	Epidermolysis bullosa simplex, Weber-Cockayne type;Epidermolysis bullosa simplex, recessive 1;Dermatopathia pigmentosa reticularis;Epidermolysis bullosa simplex, Dowling-Meara type;Naegeli-Franceschetti-Jadassohn syndrome;Epidermolysis bullosa simplex, Koebner type	AD, AR
KRT2	600194	Ichthyosis bullosa of Siemens	AD
KRT5	148040	Epidermolysis bullosa simplex-MP;Epidermolysis bullosa simplex, Koebner type;Epidermolysis bullosa simplex, recessive 1;Epidermolysis bullosa simplex, Weber-Cockayne type;Epidermolysis bullosa simplex 2B, generalized intermediate;Epidermolysis bullosa simplex 2A, generalized severe;Epidermolysis bullosa simplex-MCR;Dowling-Degos disease 1;Epidermolysis bullosa simplex 2C, localized;Epidermolysis bullosa simplex, Dowling-Meara type;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, AR	AD, AR
KRT74	608248	Woolly hair, AD;?Ectodermal dysplasia 7, hair/nail type;?Hypotrichosis 3	AD, AR
KRT85	602767	Ectodermal dysplasia 4, hair/nail type	AR

KRT9	607606	Palmoplantar keratoderma, epidermolytic	AD
LAMA3	600805	Laryngoonychocutaneous syndrome;Epidermolysis bullosa, junctional, Herlitz type;Epidermolysis bullosa, generalized atrophic benign	AR
LAMB3	150310	Epidermolysis bullosa, junctional, non-Herlitz type;Amelogenesis imperfecta, type IA;Epidermolysis bullosa, junctional, Herlitz type	AR, AD
LAMC2	150292	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa, junctional, Herlitz type	AR
LIPH	607365	Woolly hair, AR 2 with or without hypotrichosis;Hypotrichosis 7	AR
LIPN	613924	Ichthyosis, congenital, AR 8	AR
LORICRIN	152445	Vohwinkel syndrome with ichthyosis	AD
LPAR6	609239	Woolly hair, AR 1, with or without hypotrichosis;Hypotrichosis 8	AR
LRMDA	614537	Albinism, oculocutaneous, type VII	AR
LYST	606897	Chediak-Higashi syndrome	AR
MBTPS2	300294	Osteogenesis imperfecta, type XIX;?Olmsted syndrome, XL;Keratosis follicularis spinulosa decalvans, XL;IFAP syndrome with or without BRESHECK syndrome	XLR
MC1R	155555	[Skin/hair/eye pigmentation 2, blond hair/fair skin];[Skin/hair/eye pigmentation 2, red hair/fair skin];Melanoma, cutaneous malignant, 5;UV-induced skin damage;[Analgesia from kappa-opioid receptor agonist, female-specific];Albinism, oculocutaneous, type II, modifier of	AR

MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8;Waardenburg syndrome, type 2A;Waardenburg syndrome/ocular albinism, digenic;Tietz albinism-deafness syndrome;COMMAD syndrome	AD, AR
MLPH	606526	Griscelli syndrome, type 3	AR
MMP1	120353	COPD, rate of decline of lung function in;Epidermolysis bullosa dystrophica, AR, modifier of	AR
MPLKIP	609188	Trichothiodystrophy 4, nonphotosensitive	AR
MYH9	160775	Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
MYO5A	160777	Griscelli syndrome, type 1	AR
NECTIN1	600644	Orofacial cleft 7;Cleft lip/palate-ectodermal dysplasia syndrome	AR
NHP2	606470	Dyskeratosis congenita, AR 2	AR
NIPAL4	609383	Ichthyosis, congenital, AR 6	AR
NOP10	606471	Dyskeratosis congenita, AR 1	AR
NOTCH1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
NSDHL	300275	CK syndrome;CHILD syndrome	XLR, XLD
OCA2	611409	Albinism, oculocutaneous, type II;[Skin/hair/eye pigmentation 1, blue/nonblue eyes];[Skin/hair/eye pigmentation 1, blond/brown hair];Albinism, brown oculocutaneous	AR
PARN	604212	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4;Dyskeratosis congenita, AR 6	AD, AR

PAX3	606597	Waardenburg syndrome, type 3;Rhabdomyosarcoma 2, alveolar;Craniofacial-deafness-hand syndrome;Waardenburg syndrome, type 1	AD, AR, SM, AD
PEX7	601757	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PHGDH	606879	Phosphoglycerate dehydrogenase deficiency;Neu-Laxova syndrome 1	AR
PHYH	602026	Refsum disease	AR
PKP1	601975	Ectodermal dysplasia/skin fragility syndrome	AR
PLEC	601282	Epidermolysis bullosa simplex, Ogna type;Epidermolysis bullosa simplex with muscular dystrophy;?Epidermolysis bullosa simplex with nail dystrophy;Epidermolysis bullosa simplex with pyloric atresia;Muscular dystrophy, limb-girdle, AR 17	AD, AR
PNPLA1	612121	Ichthyosis, congenital, AR 10	AR
POMP	613386	Proteasome-associated autoinflammatory syndrome 2;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	AD, AR
PPOX	600923	Porphyria variegata	AD
PSAT1	610936	Neu-Laxova syndrome 2;?Phosphoserine aminotransferase deficiency	AR
PYCR1	179035	Cutis laxa, AR, type IIB;Cutis laxa, AR, type IIIB	AR
RAB27A	603868	Griscelli syndrome, type 2	AR
RBPJ	147183	Adams-Oliver syndrome 3	AD
RPL21	603636	Hypotrichosis 12	AD

RTEL1	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3;Dyskeratosis congenita, AD 4;Dyskeratosis congenita, AR 5	AD, AD, AR
SDR9C7	609769	Ichthyosis, congenital, AR 13	AR
SERPINB8	601697	Peeling skin syndrome 5	AR
SLC24A5	609802	Albinism, oculocutaneous, type VI;[Skin/hair/eye pigmentation 4, fair/dark skin]	AR
SLC27A4	604194	Ichthyosis prematurity syndrome	AR
SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	AR
SLC45A2	606202	[Skin/hair/eye pigmentation 5, black/nonblack hair];[Skin/hair/eye pigmentation 5, dark/fair skin];Albinism, oculocutaneous, type IV;[Skin/hair/eye pigmentation 5, dark/light eyes]	AR
SNAI2	602150	Waardenburg syndrome, type 2D;Piebaldism	AR, AD
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNRPE	128260	Hypotrichosis 11	AD
SOX10	602229	Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SPINK5	605010	Netherton syndrome	AR
ST14	606797	Ichthyosis, congenital, AR 11	AR
STS	300747	Ichthyosis, XL	XLR
SUMF1	607939	Multiple sulfatase deficiency	AR

TERT	187270	Melanoma, cutaneous malignant, 9;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1;Dyskeratosis congenita, AD 2;Leukemia, acute myeloid;Dyskeratosis congenita, AR 4	AD, AD, AR, AD, SM
TGM1	190195	Ichthyosis, congenital, AR 1	AR
TGM5	603805	Peeling skin syndrome 2	AR
TINF2	604319	Revesz syndrome;Dyskeratosis congenita, AD 3	AD
TYR	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin];Waardenburg syndrome/albinism, digenic;[Skin/hair/eye pigmentation 3, blue/green eyes];Melanoma, cutaneous malignant, susceptibility to, 8;Albinism, oculocutaneous, type IA;Albinism, oculocutaneous, type IB	AD, AR
TYRP1	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)];Albinism, oculocutaneous, type III	AR
UROD	613521	Porphyria, hepatoerythropoietic;Porphyria cutanea tarda	AD, AR
UROS	606938	Porphyria, congenital erythropoietic	AR
USB1	613276	Poikiloderma with neutropenia	AR
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	AR
WNT10A	606268	Odontoonychodermal dysplasia;Tooth agenesis, selective, 4;Schopf-Schulz-Passarge syndrome	AR, AD, AR
WRAP53	612661	Dyskeratosis congenita, AR 3	AR
ZMPSTE24	606480	Restrictive dermopathy, lethal;Mandibuloacral dysplasia with type B lipodystrophy	AR