Dermatology Boards Fodder: Genes to Know

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The purpose of this column is to alert dermatology residents to areas that are considered "high yield" for the mock and real boards in dermatology. This installment identifies commonly asked and highly askable factoids relating to genetic inheritance of diseases.

DISEASE	PATTERN	GENE	GENE FUNCTION
Atrichia with Papules 'Alopecia Universalis'	AR	(HR) HAIRLESS GENE	Zinc Finger
Oculocutaneous Albinism I	AR	TYROSINASE	Melanin Pathway
Oculocutaneous Albinism II	AR	P GENE	Unknown
Oculocutaneous Albinism III (Rufuos)	AR	(TRP1) TYROSINE RELATED PROTEIN	Stabilizes Tyrosinase
Alkaptonuria	AR	HOMOGENTISIC ACID OXIDASE	Phenylalanine & Tyrosine Breakdown Pathway
Hereditary Angioedema (Quinke's)	AD	(C1INH) C1 ESTERASE INHIBITOR	Inhibits first component of complement
Ataxia-Telangiectasia (Louis Bar)	AR	ATM	PI3-kinase like domain
Baere-Stevenson Syndrome		(FGFr2) FGF RECEPTOR 2	
Bannayan-Riley-Ruvalcaba	AD	PTEN	Tumor Suppressor
Bart's Syndrome	AD	(COL7A1) TYPE VII COLLAGEN	Anchoring Fibril
Gorlin Syndrom (Nevoid Basal Cell Carcinoma Syndrome)	AR	(PTCH) PATCHED	Inhibits "SMOOTHENED" signalling, this inhibition blocked by "HEDGEHOG"
Bloom's Syndrome	AR	(BLM) (RECQL3)	DNA helicase
Bruton's Agammaglobulinemia	XLR	(BTK GENE)	Tyrosine Kinase
Bullous Ichthyosiform Erythroderma (Epidermolytic Hyperkeratosis)	AD	KERATINS 1 & 10	Intermediate filament
Carney Complex (LAMB, NAME)	AD	(PRKAR1A)	Regulatory Subunit of Protein Kinase A
Chediak-Higashi Syndrome	AR	LYST	Lysosomal Transport
CHILD Syndrome	XLD	(EBP GENE) EMOPAMIL BINDING PROTEIN	Sterol Isomerase
Chronic Granulomatous Disease of Childhood	XLR (mostly)	CYTOCHROME B	NADPH-OXIDASE complex component (Respiratory Burst) needed to kill catalase positive bacteria
Citrullinemia	AD	ARGINOSUCCINATE SYTHETASE	Enzyme in urea cycle
Cockayne's Syndrome	AR	(CKN1) (ERCC6) XPB DNA HELICASE	DNA Helicase
Conradi-Hünermann Syndrome	XLD	(EBP)	Sterol Isomerase
	AR	(PEX7)	Peroxisomal gene
Cowden's Syndrome (Multiple HamartomaSyndrome)	AD	(PTEN)	Tumor Suppressor
Darrier-White Disease (Keratosis Follicularis)	AD	(SERCA2) CALCIUM ATPase2A2	Calcium Dependent ATPase
Dyskeratosis Congenita	XLR	DYSKERIN	Ribosome Assembly Chaperone
	AD	(TERC) TELOMERASE, RNA COMPONENT	Telomerase RNA Component
Dominant Dystrophic Epidermolysis Bullosa	AD	(Col7A1) Type VII COLLAGEN	Anchoring Fibril
GABEB		(BPAg2) COLLAGEN XVII (LAMB3) LAMININ	Structural Protein
Junctional EB with Pyloric Atresia	AR	INTEGRIN α6,b4	Structural
Junctional EB (EB Letalis, HERLITZ)	AR	LAMININ 5	Structural
EBS	AD	KERATINS 5 & 14	Intermediate filament
EBS with Myotonic Dystrophy	AR	PLECTIN	Structural
Hidrotic Ectodermal Dysplasia (Clouston's)	AD	CONNEXIN 30	Gap Junction Protein
Ectodermal Dysplasia with Skin Fragility	AD	PLAKOPHILIN 1	Structural
Ectod. Dyspl., Hypohidrotic (Christ-Seimens-Touraine Syndrome)	XLR	ECTODYSPLASIN	
Erythrokeratoderma Variabilis (EKV)	AD	CONNEXIN 31	Gap Junction Protein
Fabry's Disease (Angiokeratoma Corporis Diffusum)	XLR	ALPHA-GALACTOSIDASE A	Hydrolyzes Glycolipids and Glycoproteins
Familial Mediterranean Fever	AR	(MEFV) MARENSTRIN	PMN inhibitor
Farber's Disease (Lipogranulomatosis)	AR	ACID CERAMIDASE	Deficiency Leads to CERAMIDE ACCUMULATION
Gardner's Syndrome	AD	(APC)	Cleaves β-Catenin
Gaucher's Disease	AR	β-GLUCOCEREBROSIDASE	
Griscelli Syndrome	AR	(MTO5a) MYOSIN-Va	Melanosome Transport To Keratinocytes
Hailey-Hailey Disease	AD	(ATPase2C1)	Calcium Dependent ATPase
Herditary Hemorrhagic Telangiectasia (Osler-Weber-Rendu)	AD	ENDOGLIN ALK-1 GENE ACTIVIN RECEPTOR BINDING KINASE	TGF-β Binding Protein TGF-β Receptor

DISEASE	PATTERN	GENE	GENE FUNCTION
Homocystinuria	AR	CYSTATHIONE SYNTHETASE	Condensation of Homocysteine & Serine
Hunter's Syndrome	XLR	IDURONATE SULFATASE	
Hurler's Syndrome	AR	ALPHA-L-URONIDASE	
Ichthyosis, Lamellar	AR	TRANSGLUTAMINASE-1	
Ichthyosis, X-Linked	XLR	ARYL SULFATASE C	Steroid Sulfatase
Incontinentia Pigmenti	XLD	(NEMO) NF-κB essential modulator	Transcription Factor
Lesch-Nyhan Syndrome	XLR	(HGPRT)	Purine Salvage Pathway Enzyme
Lhermite-Duclos Syndrome	AR	(PTEN)	Tumor Suppressor
McCune-Albright Syndrome	None	(Gs-α)	Stimulates G protein increasing cAMP
MEN I	AD	(MEN1) MENIN GENE	Binds nuclear junD
MEN IIa and IIb	AD	(RET) RECEPTOR TYROSINE KINASE	Proto-oncogene
Menke's Kinky Hair Syndrome	XLR	MNK	Copper Transporting ATPase
Milroy's Disease (Nonne-Milroy-Meige Syndrome)	AD	(FLT-4) a.k.a (VEGFr-3)	Growth factor receptor
Monilethrix	AD	KRT hHb6 & hHb1 Type II	Intermediate filament
		HUMAN HAIR KERATINS 6 & 1	
Muir-Torre Syndrome	AD	(hMSH2)	Mismatch repair gene
Nail-Patella Syndrome	AD	LMX1B GENE	Homeobox domain transcripton factor
Naxos Disease	AD	JUNCTIONAL PLAKOGLOBIN	Structural Protein
		KERATIN 9	Intermediate Filament
Neimann-Pick Disease	AR	SPHYNGOMYELINASE	
Netherton's Syndrome	AR	SPINK5 Gene	Serine protease inhibitor
Neurofibromatosis I	AD	NF-1 (Neurofibromin)	Increases GTPase activity of ras
Neurofibromatosis II	AD	NF-2 (Schwannomin or Merlin)	
Pachyonychia Congenita	AD/AR	K6, K16, or K17	Intermediate filament
Papillon-Lefevre Syndrome	AR	CATHEPSIN C	Lysosomal protease
Peutz-Jeghers Syndrome	AD	STK11	Tumor suppressor
Phenylketonuria	AR	PHENYLALANINE HYDROXYLASE	
PIBIDS	AR	(XPD) (TFIIH) XERODERMA PIGMENTOSA D	DNA helicase
Piebaldism	AD	(C-kit)	Proto-oncogene (tyrosine kinase)
Porphyria Cutanea Tarda	AD	UROPORPHYRINOGEN DECARBOXYLASE	
Porphyria, Acute Intermittent	AD	PORPHOBILINOGEN DEAMINASE	
Porphyria, Congenital Erythropoietic (GUNTHER'S)	AR	UROPORHYRINOGEN III COSYNTHASE	
Porphyria, Heriditary Coproporphyria	AD	COPROPORHYRINOGEN OXIDASE	Mitochondrial Gene
Porphyria, Erythropoietic Protoporhyria (EPP)	AD/AR	FERROCHELATASE	Mitochondrial Gene
Porphyria, Variegate	AD	PROTOPORPHYRINOGEN OXIDASE	Mitochondrial Gene
Refsum Syndrome	AR	PHYTANOYL Co-A HYDROXYLASE	
Richner-Hanhart Syndrome	AR	TYROSINE AMINOTRANSFERASE	
Rothman-Thompson (Poikiloderma Congenital)	AR	(RECQL4) DNA HELICASE	DNA helicase
Rubenstein-Taybi Syndrome	AD	(CBP) CREB-BINDING PROTEIN	Involved in cAMP regulated gene expression
SCID Severe Combined Immunodeficiency Disease	AR	(ADA) ADENOSINE DEAMINASE	
	XLR	II-2 RECEPTOR	
Bullous Ichthyosis of Siemens	AD	KERATIN 2e	Intermediate filament
Sjögren-Larsson Syndrome	AR	FATTY ALDEHYDE DEHYDROGENASE	
Striate PPK 1	AD	DESMOGLEIN-1	Structural Protein
Striate PPK 2	AD	DESMOPLAKIN	Structural Protein
Takahara's Disease	AR	CATALASE	Bacterial Defense
Tangier Disease	AR	(CERP)	Cholesterol efflux regulatory protein
Tuberous Sclerosis	AD	(TSC1) on Chrom. #9 HAMARTIN GENE	
		(TSC2) on Chrom. #16 TUBERIN GENE	GTPase activating protein domain
Vohwinkel's	AD	LORICRIN GENE	Structural
Vohwinkel's with deafness	AD	CONNEXIN 26	Gap Junction Protein
Vorner's Syndrome	AD	KERATIN 9	Intermediate filament
Waardenburg's Syndrome	AD	(PAX3)	Transcription factor
		(MITF) (EDN3/SOX10)-with Hirschprung's	Transcription factor Endothelin
Werner's Syndrome	AR	(WRN) (ERCC) (XPB, D, and G)	DNA helicase
White Sponge Nevus	AD	KERATIN 4 & 13	Intermediate filament
Wiskott-Aldrich Syndrome	XLR	(WASP) (sialoglycoprotein)	Binds GTPase & Actin

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