بسم الله الرحمن الرحيم

MONA MAGHRABY

شبكة المعلومات الجامعية

التوثيق الالكتروني والميكروفيلم

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جامعة عين شمس

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قسم

نقسم بالله العظيم أن المادة التي تم توثيقها وتسجيلها
علي هذه الأقراص المدمجة قد أعدت دون أية تغيرات

يجب أن

تحفظ هذه الأقراص المدمجة بعيدا عن الغبار

MONA MAGHRABY

Anterior Segment Manifestations in Patients with Genetic Disorders

Thesis

Submitted For Partial Fulfillment of Master Degree

In Medical and Clinical Genetics

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M.B.B.Ch-2012

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2020

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List of abbreviations

ABHD12	Abhydrolase domain-containing protein 12
AD	Autosomal dominant
ADAMTSL4	A disintegrin and metalloproteinase with thrombospondin motifs like 4
Add	Addition
AGK	Acylglycerol kinase
AOS	Adams-Oliver syndrome
AR	Autosomal recessive
ARHGAP31	Rho GTPase activating protein 31
ASD	Atrial septal defect
B3GAT3	Beta-1,3-glucuronyltransferase 3
B4GALNT1	Beta-1,4-N-Acetylgalactosaminyltransferase 1
BFSP1	Beaded filament structural protein 1
BFSP2	Beaded filament structural protein 2
CDPD	Corneal dystrophy and perceptive deafness
CFC	Cardiofaciocutaneous syndrome
CGH	Comparative genomic hybridization
CHED	Congenital Hereditary Endothelial Dystrophy
CHRDL1	Chordin like 1
CIPA	Congenital insensitivity to pain with anhidrosis
CRYAA	Crystallin alpha A
CRYAB	Crystallin alpha B
CRYBA	crystallin beta A
CRYBB	crystallin beta B
CRYGA	crystalline gamma A
CRYGB	crystalline gamma B
CRYGC	crystalline gamma C
CRYGD	crystalline gamma D
CRYGS	crystalline gamma S

del	Deletion
der	Derivative chromosome
DLL4	delta like canonical Notch ligand 4
DOCK6	Dedicator of cytokinesis 6
dup	duplication
ECHO	Echocardiography
ELOVL4	Elongation of very long chain fatty acids-like 4
ELP4	Elongator acetyltransferase complex subunit 4
EOGT	EGF domain specific O-linked N-acetylglucosamine transferase
EPHA2	Ephrin receptor A2
ERG	Electroretinography
FBN	Fibrillin
FISH	Fluorescence in situ hybridization
FOXE3	Forkhead Box E3
G6PD	Glucose-6-phosphate dehydrogenase
GALT	Galactose-1-phosphate uridylyltransferase
GEMSS	Glaucoma-lens ectopia-microspherophakia-stiffness-shortness
GJA3	Gap junction protein alpha 3
GJA8	Gap junction protein alpha 8
GJB2	Gap junction protein beta 2
HCCS	Holocytochrome C synthase
HDAC9	Histone deacetylase 1
IEM	Inborn error of metabolism
inv	Inversion
IPJ	Interphalangeal joint
Lt	Left
LTBP2	latent transforming growth factor beta binding protein 2
MAPK	Mitogen-activated protein kinase
mar	Marker chromosome

MGC 1	Congenital megalocornea
MIP	Major intrinsic protein of lens fiber
MPJ	Metacarpophalangeal joint
MPS	Mucopolysaccharidosis
MR	Mitral regurgitation
MRCS	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma
MSP1	Microsperophakia 1
NOTCH1	Notch receptor 1
Nuc ish CEP X x1,SRY x1	One copy of the locus CEP X and SRY probe are observed by FISH
OAT	Ornithine aminotransferase
OCRL1	OCRL inositol polyphosphate-5-phosphatase
P5C synthetase	Pyrroline-5-carboxylate synthase
PAUS	Abdominopelvic ultrasonography
PAX6	Paired box 6
PC	Pachyonychia congenita
PDA	Patent ductus arteriosus
PEX 7	Peroxisome biogenesis factor 7
PFO	Patent foramen ovale
PPT1	Palmitoyl-protein thioesterase 1
RAX	Retina and anterior neural fold homeobox
RBPJ	Recombination signal binding protein for immunoglobulin kappa J region
RCDP	Rhizomelic chondrodysplasia punctata
ROP	Retinopathy o prematurity
Rt	Right
SLC33A1	Solute carrier family 33- member 1, Acetyl CoA transporter
SLC4A11	Sodium bicarbonate transporter-like solute carrier family 4 member 11
SNHL	Sensorineural hearing loss

SRD5A3	Steroid 5-α-Reductase 3
TGFβ2	Transforming growth factor β2
TR	Tricuspid regurgitation
TRIM44	Tripartite motif containing 44
U/S	Ultrasonography
VEP	Visual evoked potential
VSD	Ventricular septal defect
WAGR	Wilm's tumor, bilateral sporadic aniridia, genitourinary abnormalities and mental retardation
XL-D	X linked dominant
XL-R	X linked recessive

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