



شبكة المعلومات الجامعية
التوثيق الإلكتروني والميكروفيلم

بسم الله الرحمن الرحيم



MONA MAGHRABY



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شبكة المعلومات الجامعية التوثيق الإلكتروني والميكروفيلم



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جامعة عين شمس التوثيق الإلكتروني والميكروفيلم

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تحفظ هذه الأقراص المدمجة بعيدا عن الغبار



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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
CHRD1	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	Pyroline-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 of 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
TGF β 2	Transforming growth factor β 2
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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