Study of Some Egyptian Patients with Multiple Congenital Anomalies

Thesis

Submitted for Partial Fulfillment of the Requirements of Master Degree of Genetics

By

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دراسة لبعض المرضى المصريين اللذين يعانون من التشوهات الخلقية متعددة

رسالـــة

توطئة للحصول على درجة ماجستير في طب الوراثة

مقدمة من

الطبيبة/ هبه أمجد عبد الحميد

بكالوريوس في الطب والجراحة جامعة عين شمس (٢٠١٠)

تحت إشراف

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أستاذ طب الأطفال وعلم الوراثة

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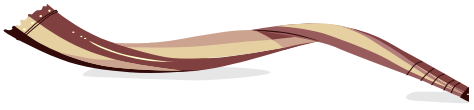
Heba Amgad



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List of Abbreviations

Abb.	Meaning
AARR	Al Awadi Raas Rothschild
aCGH	Array comparative genomic hyperidization
ARC	Arthrogryposis, renal tubular acidosis and cholestasis
ARO	Autosomal recessive osteopetrosis
ASD	Atrial septal defect
ASD	Autism spectrum disorder
B19V	Parvovirus B19
BMP	Bone morphogenicprotien
CD	Campomelic dysplasia
CFD	Congenital fibrogenemia disorder
CHD	Congenital heart disease
CHILD	Congeniatl Hemidysplasia, Ichthyosiform erythroderma and limb defects
CLOVE	Congenital Lipomatous Overgrowth, Vascular malformation, Epidermal nevi
CMA	Chromosomal microarray analysis
CMs	Congenital malformations
CNS	Central nervous system
CRL	Crown rump length
CSF	Cerebrospinal fluids
CVS	Chrionic villus sapling
DHODH	Dihydroorotate dehydrogenase
EMG and NCV	Electromyography and nerve conduction velocity

Abb.	Meaning
FA	Folic acid
FISH	Florescent in situ hybridization
FSS	Freeman Sheldon syndrome
GMW	Galaway Mowat syndrome
HCG	Human chorionic gonadotrophin
HIV	Human immunodeficiency virus
HSV	Herpes simplex virus
ICCG	International collaboration for clinical genomics
ICD 10	International classification of disease
ID/DD	Intellectual disabilities / developmental delay
INH	Isoniazide
IOM'S	Institute of medicine
ISH	Infantile systemic hyalanosis
JHF	Juvenile hyaline fibromatosis
JSRD	Joubert syndrome and related disorders
LCMV	Lymphocytic chorio meningitis virus
LWD	Leri Weill dyschondrostosis
MCA	Multiple congenital anomalies
MDS	Myelodysplastic changes
MLPA	Multiplex ligation- dependent probe amplification
MSAFP	Maternal serum alpha fetoprotien
MTS	Molar tooth sign
NICU	Neonatal intensive care unit
NTDs	Neural tube defect

Abb.	Meaning
OFDS VI	Oro faciodigital type VI
PAS	Para aminosalycilic acid
PCR	Polymerase chain reaction
PDA	Patent ductus arteriosus
PFFD	Proximal focal femoral deficiency
PFO	Patent foramen oval
PUBS	Percutaneous umblical blood sapling
RDA	Recommended daily allowance
SHOX	Short stature home box
SNP	Single nucleotide polymorphism
SVAS	Supra valvular aortic stenosis
TAR	Thrombocytopenia absent radius syndrome
TORCH	Toxoplasma, others, rubella, cytomegalovirus, herpes simplex
UPJA	Uretopelvi junction obstruction
VEP	Visual evoked potential
VPI	Velopharyngeal incompetence
VSD	Ventricular septal defect
VXs	Verruciform xanthomas
VZV	Varicella zoster virus
WHS	Wolf Hirschhorn syndrome

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