Introduction

Adoption

Approach to the consultation with a child with dysmorphism, congenital malformation or developmental delay

Autosomal dominant (AD) inheritance

Autosomal recessive (AR) inheritance

Communication skills

Complex inheritance

Confidentiality

Confirmation of diagnosis

Consent for genetic testing

Genetic basis of cancer

Genetic code and mutations

Genomes and genomic variation

Genomic imprinting

Genomic sequencing and interpretation of data from WES or WGS analyses

Mitochondrial inheritance

Reproductive options

Testing for genetic status

Timing and origin of new dominant mutations

Useful resources

X-linked dominant (XLD), semi-dominant, pseudoautosomal and male sparing inheritance

X-linked recessive inheritance

Clinical Approach

Ambiguous genitalia (including sex reversal)

Anal anomalies (atresia, stenosis)

Anterior segment eye malformations

Arthrogryposis

Ataxic adult

Ataxic child

Brachydactyly

Broad thumbs

Cardiomyopathy in children under 10 years

Cataract

Cerebellar anomalies

Cerebral palsy

Chondrodysplasia punctata

Cleft lip and palate

Coarse facial features

Coloboma

Congenital heart disease

Congenital hypothyroidism

Corneal clouding

Deafness in early childhood

Developmental delay in the child with consanguineous parents

Developmental regression
Duane retraction syndrome
Dysmorphic child
Dystonia
Ear anomalies
Facial asymmetry
Failure to thrive
Floppy infant
Fractures
Generalized disorders of skin pigmentation (including albinism)
Hemihypertrophy and limb asymmetry
Holoprosencephaly
Hydrocephalus
Hypermobile joints
Hypoglycaemia in the neonate and infant
Hypospadias
Intellectual disability
Intellectual disability with apparent X-linked inheritance
Increased bone density
Intracranial calcification
Large fontanelle
Laterality disorders including heterotaxy and isomerism
Leukodystrophy/leukoencephalopathy
Limb reduction defects
Lissencephaly, polymicrogyria and neuronal migration disorders
Lumps and bumps
Macrocephaly
Microcephaly
Micrognathia and Robin sequence
Microphthalmia and anophthalmia
Minor congenital anomalies
Nasal anomalies
Neonatal encephalopathy and intractable seizures
Nystagmus
Obesity with and without developmental delay
Ocular hypertelorism
Oedema generalized or puffy extremeties
Oesophageal and intestinal atresia (including tracheo-oesophageal fistula)
Optic nerve hypoplasia
Overgrowth
Patchy hypo- or de-pigmented skin lesions
Patchy pigmented skin lesions (including café-au-lait spots)
Plagiocephaly and abnormalities of skull shape
Polydactyly
Prolonged neonatal jaundice and jaundice in infants below 6 months

Ptosis, blepharophimosis and other eyelid anomalies
Radial ray defects and thumb hypoplasia
Retinal dysplasia
Retinal receptor dystrophies
Scalp defects
Seizures with developmental delay/intellectual disability
Short stature
Skeletal dysplasias
Structural intracranial anomalies (agenesis of the corpus callosum, septo-optic dysplasia and arachnoid cysts)
Sudden cardiac death
Suspected non-accidental injury
Syndactyly (other than 2-3 toe syndactyly)
Unusual hair, teeth, nails and skin

Common consultations

Achondroplasia
Alpha1-antitrypsin deficiency
Alport syndrome
Androgen insensitivity syndrome (AIS)
Angelman syndrome
Autism and autism spectrum disorders
Autosomal dominant polycystic kidney disease (ADPKD)
Beckwith-Wiedemann syndrome (BWS)
Charcot-Marie-Tooth disorder (CMT)
Ciliopathies
Congenital adrenal hyperplasia (CAH)
Consanguinity
Craniosynostosis
Cystic fibrosis (CF)
Dementia early onset and familial forms
Diabetes mellitus
Dilated cardiomyopathy (DCM)
DNA repair defects
Duchenne and Becker muscular dystrophy (DMD and BMD)
Ehlers-Danlos syndrome
Epilepsy in infants and children
Epilepsy in adults
Fascioscapulo-humeral muscular dystrophy (FSHD)
Fragile X syndrome (FRAX)
Glaucoma
Haemochromatosis
Haemoglobinopathies
Haemophilia and other inherited coagulation disorders
Hereditary haemorrhagic telangiectasia (HHT)
Herediatry spastic paraplegia (HSP)

Hirschprung disease
Huntington disease (HD)
Hyperlipidaemias
Hypertrophic cardiomyopathy (HCM)
Immunodeficiency and recurrent infection
Incest
Leigh encephalopathy
Limb-girdle muscular dystrophies
Long QT and Brugada syndromes
Marfan syndrome
Mitochondrial DNA diseases
Myotonic dystrophy (DM1)
Neural tube defects
Neurofibromatosis type 1 (NF1)
Noonan syndrome and the RAS-MAPK pathway disorders
Parkinson disease
Retinitis pigmentosa (RP)
Rett syndrome
Sensitivity to anaesthetic agents
Spinal muscular atrophy (SMA)
Stickler syndrome
Thrombophilia
Tuberous sclerosis (TSC)
X-linked adrenoleukodystrophy (X-ALD)

Cancer

BRCA1 and BRCA2
Breast cancer
Cancer surveillance methods
Colorectal cancer (CRC)
Confirmation of diagnosis of cancer
Cowden syndrome (CS)
Familial Adenomatous Polyposis (FAP) and adenomatous polyposis (due to MUTYH, NTHL1, POLE & POLD1)
Gastric cancer
Gorlin syndrome
Juvenile polyposis syndrome (JPS)
Lynch syndrome
Lifestyle factors in cancer: smoking, alcohol, obesity, diet and exercise
Li-Fraumini syndrome (LFS)
Multiple endocrine neoplasia (MEN)
Neurofibromatosis type 2 (NF2)
Ovarian cancer
Peutz-Jeghers syndrome (PJS)
Phaeochromocytoma and Paraganglioma
Prostate cancer

Renal cancer
Retinoblastoma
von Hippel-Lindau (VHL) disease
Wilms tumour

Chromosomes

22q11 deletion syndrome
47,XXX
47,XXY
47,XYY
Autosomal reciprocal tranlsocations background
Autosomal reciprocal translocations familial
Autosomal reciprocal translocations postnatal
Autosomal reciprocal translocations prenatal
Cell division mitosis, meiosis and non-disjunction
Chromosomal mosaicism postnatal
Chromosomal mosaicism prenatal
Deletions and duplications (including microdeletions and microduplications)
Down syndrome (trisomy 21)
Edwards syndrome (trisomy 18)
Inversions
Mosaic trisomy 8
Mosaic trisomy 16
Patau syndrome (trisomy 13)
Prenatal diagnosis of sex chromosome aneuploidy
Ring chromosomes
Robertsonian translocations
Sex chromosome mosaicism
Supernumerary marker chromosomes (SMCs) postnatal
Supernumerary marker chromosomes (SMCs) prenatal
Triploidy (69,XXX, 69XXY or 69,XYY)
Turner syndrome, 45,X and variants
X-autosome translocations

Pregnancy and fertility

Anterior abdominal wall defects
Assisted reproductive technology: in vitro fertilization (IVF), intracytoplasmic sperm injection (ICSI) and pre-implantation genetic diagnosis (PGD)
Bowed limbs
Congenital cystic lung lesions, Currarino syndrome, and sacrococcygeal teratoma
Congenital diaphragmatic hernia
Cytomegalovirus (CMV)
Drugs in pregnancy
Female infertility and amenorrhoea: genetic aspects
Fetal alcohol syndrome (FAS)
Fetal anticonvulsant syndrome (FACS)
Fetal akinesia

Fetomaternal alloimmunisation (rhesus D and thrombocytopaenia)
Hyperechogenic bowel
Hyoplastic left heart
Imaging in prenatal diagnosis
Invasive techniques and genetic tests in prenatal diagnosis
Low maternal serum oestriol
Male infertility: genetic aspects
Maternal age
Maternal diabetes mellitus and diabetic embryopathy
Maternal phenylketonuria (PKU)
Miscarriage and recurrent miscarriage
Neonatal (newborn) screening (NS)
Non-invasive prenatal diagnosis/testing (NIPD/T)
Oedema increased nuchal translucency, cystic hygroma and hydrops
Oligohydramnios (Including Potter/ Oligohydramnios sequence)
Paternal age
Polyhydramnios
Posterior fossa malformations
Premature ovarian failure (POF)
Radiation exposure, chemotherapy, and landfill site
Rubella
Short limbs
Talipes (Club foot)
Toxoplasmosis
Twins and twinning
Urinary tract and renal anomalies (Congenital anomalies of the kidney and urinary tract - CAKUT)
Varicella
Ventriculomegaly

Appendix

Antenatal and neonatal screening timelines
Bayes theorem
Carrier frequency and carrier testing for autosomal recessive disorders
Centile charts for boys height and weight
Centile charts for girls height and weight
Centile charts for occipital-frontal circumference (OFC)
CK (Creatine kinase) levels in carriers of Duchenne muscular dystrophy (DMD)
Conversion charts from English to metric units for height and weight
Denver Developmental Screening Test
Distribution of muscle weakness in different types of muscular dystrophy
Dysmorphology examination checklist
Embryonic fetal development (overview)
Family tree sheet and symbols
Haploid autosomal lengths of human chromosomes
Investigation of lethal metabolic disorder or skeletal dysplasia

ISCN Nomenclature
Karyotypes
Normal range of aortic root dimensions
Paternity testing
Patterns of cancer
Radiological investigations including magnetic resonance imaging (MRI)
Skeletal dysplasia charts
Staging of puberty