Part	Sections	# of pages	Chapters	COMPULSORY subchapters (perhaps one author per subchapter)	SUGGESTED topics/subchapters to cover Some may become subchapters with co-author	Links to clinical vignettes (may be > 1)
Part 1. HUMAN GENETICS						
	S1. Introduction to Human Genetics					
		5	**Chapter 1-1. Introduction: genetics and medicine**		Medical genetics Genetic diseases The heterogeneity of genetic diseases Rare diseases	
	S2. Genomics					
		10	**Chapter 2-1. Basics of nucleic acids biology**		DNA biochemistry, The genetic code Organization of human genes Coding and functional,non-coding RNAs, pseudogenes, repeated DNA elements, Organization of human genome and of the chromatin	
		10	**Chapter 2-2. Regulation of gene transcription**		Mechanism of transcription The histone code and epigenetic regulation of the genome Regulation of the transcription / no-coding RNAs	
		5	**Chapter 2-3. Regulation of gene translation**		Mechanism of translation Post-translational modifications	
		10	**Chapter 2-4. Mitosis and meiosis**		Cell cycle and its regulation, DNA replication and chromosome formation Mitosis, Méiosis	
		5	**Chapter 2-5. Transmission of the genome**		Transmission of the genome & gametogenesis	
	S3. Variants et mutations					
		15	**Chapter 3-1. Qualitative and quantitative variations of the genome**		General description of normal variation, irrespective of the potential pathogenic effects : MS, NS, Splicing, ESE/ESS, deep intronic, promotor 5'UTR, INDELs, , dynamic variations triplet, DMPK...) Structural genomic rearrangements, balanced and unimbalanced Anomaly in the number of chromosomes Mosaicism UPD	

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		10	**Chapter 3-2. Functional consequences of mutations on proteins (selected examples)**		Discussion of LoF/hypomorpg, GoF, hypermorph, néomorph, antimorph, dominant neg and other functional effects. Enzymes, Receptors, Channels, Signal transduction, Structural proteins Other pathogenic mechanism: modification of ncRNA, miRNA...	
		10	**Chapter 3-3. Formal genetics: heredity modes**		AD, AR, XLR, Pseudoautosomal heredity Chromosomal inheritance Mitochondrial inheritance Non-mendelian inheritance Mosaicism Uniparental disomy	
		5	**Chapter 3-4. Genotype/phenotype relationships**		Functional appoach to Mendian inheritance: basis of dominant and of recessive inheritance at cellular level (distinct from the "formal" and classical aspect in section on clinical gzetics	
		5	**Chapter 3-5. Mechanism for the emergence of mutations**		Basics of point mutation, gene conversion, Including genomic variation: Low copy repeats, NAHR, FOSTES	
		5	**Chapter 3-6. Descriptive nomenclature of gene and genomic anomalies**		Descriptive nomenclature of gene anomalies (HGVS) Descriptive nomenclature of cytogenomic anomalies (ISCN)	
		5	**Chapter 3-7. Strategies to link a phenotype to a gene**		Genetic linkage studies in families positional cloning, genetic map, recombination rate LOD-Score	
		5	**Chapter 3-8. Transgenesis**		Additive transgenesis and homologous recombination in ES cells: KO, KI, conditional KO (Cre-LoxP) Gene editing CRISPR-Cas9	
		5	**Chapter 3-9. Model organisms**		General overview o f the main model organisms	
	S4. Multifactorial/genetic predisposition to common diseases					

NB: One A4 page (12pt) ≈ 3000ch (incl. spaces)

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		15	**Chapter 4-1. Multifactorial/genetic predisposition to common diseases**		Complex diseases Heritability Common alleles or rare alleles, continuous polygenic model and polygenic threshold model Linkage analysis Association studies Pangenomic association studies	
	S5. Genetics and human populations					
		10	**Chapter 5-1. Genetics and human populations**		Hardy and Weinberg equilibrium Linkage and linkage desequilibrium Disturbing HW equilibrium Stratification of populations Migration, genetic flux and genetic drift Preferential unions, endogamy and consanguinity Mutation and natural selection Human races do not exist	
Part 2. CLINICAL APPLICATIONS OF GENETICS						
	S6. Omics					
		10	**Chapter 6-1. Classical technologies**	**Classical cytogenetics and FISH**	Description of the main techniques	
				Molecular	Historical aspects: positional cloning of disease-causing genes Detection of short sequence variants PCR Sanger sequencing Expansion detection Methylation detection Quantitative dosages	
		10	**Chapter 6-2. Genomics technologies**	**Molecular cytogenetics**	CGH and SNP arrays	
				NGS	NGS technologies (short reads) NGS technologies (long reads)	
				Other	Detecting structural rearrangements (Nano…)	
		3	**Chapter 6-3. Transcriptomics technologies**		RNA seq Gene expression profiling	
		5	**Chapter 6-4. Epigenomics technologies**		Investigation of chromatine architecture ATAC-Seq Conformation capture technologies	
		2	**Chapter 6-5. Proteomics and metabolomics technologies**		Mass spectrometry	
		5	**Chapter 6-6. Establish the pathogenicity of a variant**		ACMG classification	
	S7. Clinical genetics and genetic counseling					

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		5	**Chapter 7-1. Basics of risk calculation applied to genetics**		Risk calculation Bayes	
		10	**Chapter 7-2. Genetic counseling**		Pedigree drawing Genetic counseling in Mendelian disorders Genetic counseling in genomic disorders Genetic counseling in common disorders Genetic clinics : practical aspects /communication strategy /…	
		5	**Chapter 7-3. Carrier testing in families**			
		5	**Chapter 7-4. Presymptomatic diagnosis**			
		3	**Chapter 7-5. Regulatory aspects of the practice of genetic testing in EU**		Regulatory aspects of the practice of genetic testing in EU (overview + annex: 27 sections by country)	
		3	**Chapter 7-6. The management of rare diseases in EU and the ERNs**		The management of rare diseases in EU and the ERNs	
	S8. Genetic control of normal human development					
		5	**Chapter 8-1. Developmental genetics**	**Overview**	Genetic control of development Epigenetic control of development (examples by organ are in the pathway section)	
		2	**Chapter 8-2. Genetic of developmental pathways**	**DP1: Early embryonic development and Left-Right Asymmetry**	Basics of embryologic development with the main gene partners (may refer to pathways and processes described in the next sections: S9 to S13	**May refer to some of the clinical vignettes or to clinically oreinted prorblms (discussed in part 4)**
		2		**DP2: Neural Crest Formation and Craniofacial Development**	Idem	**Idem**
		2		**DP3: Development of the Nervous System: neural tube**	Idem	**Idem**
		2		**DP4: Development of the Nervous System: neuronogenesis**	Idem	**Idem**
		2		**DP5. Development of the Eye**	Idem	**Idem**
		2		**DP6. Development of the Ear**	Idem	**Idem**
		2		**DP7. Muscle and Somite Development**	Idem	**Idem**
		2		**DP8. Bone and Cartilage**	Idem	**Idem**
		2		**DP9. Development of the limbs**	Idem	**Idem**
		2		**DP10. Cardiogenesis**	Idem	**Idem**
		2		**DP11. Kidney**	Idem	**Idem**

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		2		DP12. Development of the Endodermal Derivatives: Lung, Liver, Pancreas, and Gut	Idem	Idem
		2		DP13. Development of skin and Epidermal Appendages: Teeth and Hair	Idem	Idem
		2		DP14. Control of sex determination	Idem	Idem
Part 3. GENETICS IN MEDICINE : etiopathogenic approach						
	S9. Genetic basis of metabolic processes					
		2	Chapter 9-1. Metabolic disorders (inborn errors)	A1. amino acid metabolism & urea cycle	Organisation of the chapter to be defined by JZ : current sections with A# used for cross indexing with Vignettes, but may be merged	
		2		A2. carbohydrate metabolism		
		2		A3. fatty acid, carnitine and ketone body		
		2		A4. Mitochondria: energy metabolism		
		2		A5. lipid metabolism		
		2		A6. lipoprotein metabolism		
		2		A7. Complex molecule degradation – lysosomal disorders		
		2		A8. Peroxisomes		
		2		A9. purine, pyrimidine and nucleotide metabolism		
		2		A10. Cofactors ands vitamins		
		2		A11. Porphirins		
		2		A12. Steroids		
		2		A13. Glycosylation (O and N)		
		2		A14. Metal		
	S10. Genetic basis of Intracellular signalling					
		2	Chapter 10-1. Signaling pathways	Cell receptors overview	General principles of cell signaling	
		2		C1. SHH Signaling Pathway	Overview of the pathway with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
		2		C2. Wnt Signaling Pathway	Idem	
		2		C3. TGF-b Signaling Pathway	Idem	
		2		C4. FGF Signaling Pathway	Idem	
		2		C5. GDNF Signaling Pathway	Idem	
		2		C6. RTK/RAS/MAP & PI3K/AKT pathways	Idem	

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		2		C7. I: EPHRIN Signaling Pathway	Idem	
		2		C8. TNF Signaling pathway	Idem	
		2		C9. NOG pathway	Idem	
		2		C10. MYC pathway	Idem	
		2		C11. Notch pathway	Idem	
		2		C12. p53 pathway	Idem	
		2		C13. Guanine nucleotide Binding protein	Idem	
		2	Chapter 10-2. Transcription factors	D1. The Homeobox Gene Family	Idem	
		2		D2. The Paired Box (PAX) Gene Family	Idem	
		2		D3. The Forkhead Gene Family	Idem	
		2		D4. The T-Box Gene Family	Idem	
		2		D5. THE SOX gene family	Idem	
		2		D6: other TF	Idem	
		2	Chapter 10-3. Regulation of chromatin function	E1. Histone/DNA méthylation regulation	Idem	
		2		E2. Cohesines	Idem	
		2	Chapter 10-4. Regulation of protein synthesis	F1. A: RNA processing (localisation, spliceosome)	Idem	
		2		F2. Post-translational control & ubiquitinisation	Idem	
	S11. Genetic basis of cell dynamics					
		2	Chapter 11-1. Cellular dynamics	F3. Cell cycle and apoptosis	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
		4		F4. DNA repair mechanisms	Idem	
		2		F5. Cytoskeletton	idem	
		2		F6. Vesicle-mediated trafficking	Idem	
	S12. Genetic basis of Extracellular matrix					
		2	Chapter 12-1. Extracellular matrix	G1. Collagen & fibrillin	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
	S13. Genetic basis of Junctions, transporters and channels					
		8	Chapter 13-1. Junctions, transporters and channels	H1. GAP junctions	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
				H2. Channels	Idem	

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				H3. Membrane transporter	Idem	
				H4. Membrane proteins	Idem	
	S14. Genetic basis of Ciliary Functions: Genesis, Transport, and Reabsorbtion					
		3	**Chapter 14-1. Ciliary Functions: Genesis, Transport, and Reabsorbtion AND mitile cilia**	**B1. Primary cilium and its function**	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
	S15. Genetic basis of red cell metabolism					
		4	**Chapter 15-1. Red cell metabolism**	**I1. Heme synthesis**	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
				I2. Hemoglobin synthesis	Idem	
	S16. Genetic basis of Hemostasis					
		3	**Chapter 16-1. Hemostasis**	**J1. Coagulation cascade**	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
				J2. Platelet function	Idem	
	S17. Genetic basis of Immune system					
		3	**Chapter 17-1. Immune system**	**K1. Ig and T cell receptors**	Overview of the cellular process with visual representation of the pathway, overview of the major physiological processes, and a table or list of the disorders that involve this pathway	
Part 4. GENETICS IN MEDICINE : clinically oriented approach						
	S18. Dysmorphology and congenital malformations					
		5	**Chapter 18-1. Congenital malformations:**		Congenital malformations: epidemiology & classification	
		5	**Chapter 18-2. Dysmorphology**	**Basics of dysmorphology**	Examination of a child with MCA or dysmorphic features	
		2	**Chapter 18-3. Craniofacial anomalies**	**Abnormal skull shape**	Clinical approach of a patient with Craniosynostosis (basics of classification, diagnotic process)	**Craniofrontonasal Syndrome and EFNB/ Mutations 87** **Craniosynostosis 61** **FGF Receptor Mutations**
		2		**Facial clefting**	Clinical approach to a child with Cleft or Robin sequence	
		2		**Abnormal ear shape**	clinical approch to a child with morphological ear anomaly	**Treacher-Collins. TCOF1**

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		2		**Oculopalpebral anomalies**	Oculopalpebral anomalies	**Frontonasal Dysplasias 104. The ALX Homeobox Gene Family** **BPES**
		2	**Chapter 18-4. Selected malformations**	**Limb malformation**	General clinical overview and classification of limb anomalies (refer to the section 3 for embryology)	**Holt-Oram/Okihiro Syndromes 121. TBX3, TBX5, and the Ulnar-Mammary and**
		2		**Congenital heart disorders**	Clinical approach to a child with CHD	
		2		**Neural tube defects**	Clinical approach to a child with dysraphism	**Currarino Syndrome 99. HLXB9 (MNX1!). Sacral Agenesis. and the**
		2		**Multiple congenital anomalies (selected examples)**	PROVISIONAL Clinical approach to a child with multiple birth defects (may be dispatched to more specific entries)	**Bardet-Biedl** **Beckwith-Wiedemann** **CHARGE 148. CHD7 and** **Cohen** **Cornelia de Lange** **Del 22q11.2, Del 4p, Del 5p** **Down syndrome** **Pallister-Hall and Greig Syndromes 32. GLI3 and the** **RA1, Smith-Magenis Syndrome, and** **Potocki-Lupski Syndrome 134. Rubinstein-Taybi Syndrome 141. CREBBP** **Silver-Russell** **TAR** **Triploidy** **Trisomy 13** **Trisomy 18** **VACTERL** **Williams**
S19. Environmentallly-induced birth defects						
		2	**Chapter 19-1. Chemical exposure**		General overview of major teratogenic agents	**Anticonvulsant therapy** **Fetal Alcohol exposure**
		2	**Chapter 19-2. Infections**		General overview of major infectious agents	**Rubella, toxo, CMV, Zika**
		4	**Chapter 19-3. Irradiation**		Effect of irradiations	
S20. Genetics of neurodevelopment, including ID and ASD						
		10	**Chapter 20-1. Genetic and neurodevelopment, including ID and ASD**	**Neurodevelopmental anomalies: overview**	Medical definition of intelligence Definition of ID, ASD and the NDD Etiologies and epidemiology of ID and ASD Medical care (bases)	
				Genetic bases of NDD	General overview of etiologies of NDD	
				Intellectual disability and NDD: evaluation et diagnostic strategies	Clinical approach to a child with a NDD	

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				Syndromal ID syndromes	PROVISIONAL Clinical approach to a chold with multiple birth defects (may be dispatched to more specific entries)	**Carbohydrate Deficient Glycosylation (N-glycosylation)** **Coffin-Lowry Syndrome 146. RSK2 and the** **Cohen Syndrome 192. VPS/3B and Del/dup 16p11.2** **Fragile X Syndrome 158. FMRI and the** **Kabuki Syndrome 153.** **Phenylketonuria:** **Rett syndrome 520** **Smith-Lemli-Opitz Syndrome** **Sotos syndrome (NDS1)** **Maternal UPD 14**
	S21. Genetics of Central and peripheral nervous system					
		5	**Chapter 21-1. Brain malformations (EPICARE)**	**Clinical approach to a child with structural brain anomaly**	General overview of Holoprosencephaly and hydrocephaly Clinical approach to a child with a Structural brain defect, including agenesis of the CC	**Feingold Syndrome 34. MYCN and Microcephaly (ASPM)** **Microcephaly + dwarfism (MOPD2)** **Lissencephaly (LIS)** **Periventricular Nodular Heterotopia & OPD FLNA and FLNB** **Walker-Warburg Syndrome: Abnormal Glycosylation of Alpha-Dystroglycan 206.** **Joubert Syndrome and Related Disorders 20. The Molecular Basis of LICAM syndrome** **Joubert Syndrome and Related Disorders 20**
		15	**Chapter 21-2. Neurological disorders (EPICARE + RMD)**		General overview of Epilepsy and its approach	
				Clinical approach to a child with hypotonia **Clinical approach to a patient with a Spastic paraplegia**		**Hereditary spastic paraplegias (HSP) 448**

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				Clinical approach to a patient with ataxia or another movement disorder		**Ataxia-telangiectasia ATM, MRE11A** **Friedreich ataxia**
				Clinical approach to a child with Neurodegenerative disorder	General overview of neurdegenerative disorder in children	**Canavan** **Leigh** **Lipofuscinoses** **Menkes** **Pelizaeus** **Tay-Sachs disease:** **X-linked adrenoleukodystrophy (X-ALD) 538** **Rett**
				Clinical approach to an adult patient with Neurodegenerative disorders	General overview of neurdegenerative disorders in adults	**Dementia—early onset and familial forms 386** **Huntington's disease:** **Parkinson**
				Clinical approach to a patient with a Neurovascular diseases	General overview of neurovaculardisorders in adults	**Cerebral aneurysms** **Cerebral Cavernous Malformations 219. KRIT!, CCM2, and PDCD/O**
				Clinical approach to a patient with a Neuropathy	General overview of neurovaculardisorders in adults	**Amyotrophic lateral sclerosis** **Charcot-Marie-Tooth disease (CMT) 362** **Spinal muscular atrophy (SMA) 526**
		5	**Chapter 21-3. Neuromuscular disorders (NMD)**	**Clinical approach to a patient with a muscle disorders**	General overview of myopathies and muscular dystrophies, aand clinical approach	**"Carnitine uptake defect** **Myastenia gravis** **Pompe disease"** **"Duchenne and Becker muscular dystrophy (DM D and BMD) 402** **Facioscapulohumeral muscular dystrophy (FSHD) 420** **Steinert Myotonic dystrophy (DM1) 496** **Malignant hyperthermia"** **Myastenia gravis** **Pompe disease**
				Clinical approach to a child with Arthrogryposis multiplex	General overview of arthrogryposis disorder in children	**Escobar**
S22. Genetics of Heart and vessels disorders						
		3	**Chapter 22-1. Heart disorders HEART)**	**Clinical approach to a patient with a cardiomyopathy**		**Hypertrophic cardiomyopathy (HCM) 460** **Dilated cardiomyopathy**

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				Clinical approach to a patient with a conduction disorder		Long QT syndrome and other inhented arrhythmia syndromes 480
		3	Chapter 22-2. Vascular disorders (VASCERN)	Clinical approach to a patient with a disorder of large vessels	General overview of disorders of large and smal vessels	Marfan, Ehlers Danlos IV, aortic aneurysm, Rendu-Osler
	S23. Genetics of Immuno-hematological disorders					
		3	Chapter 23-1. Coagulation disorders (EuroBloodNet)	Clinical approach to a patient with an Hemolysis	General overview of hemolytic anaemias	G6PD- glucose 6 phosphate dehydrogenase
				Clinical approach to a patient with a Coagulation defect	General overview of coagulation defects	Haemophilia and other inherited coagulation disorders 442 Thrombophilia 530
		3	Chapter 23-2. Hemoglobin disorders (EuroBloodNet)		General overview of Hb pathies	Sickle cell anaemia: Thalassemia, alpha & beta
		3	Chapter 23-3. Immune disorders (RITA)		B cell immunodeficiencies T cell Immunodeficiencies SCID	Bruton agammaglobulinemia
	S24. Genetics of Visceral disorders					
		2	Chapter 24-1. Pulmonary disorders (Lung)	Cystic fibrosis	Overview of CF	Cystic fibrosis (CF) 382
		4	Chapter 24-2. Liver, pancreas and digestive tract disorders LVER - partim + ?)	Clinical approach to a patient with an intestinal disorder		Hirschprung (RET) Mowat- Wilson Syndrome 54. ZEB
				Clinical approach to a patient with a Liver disorder		Acute intermmittent porphyria Allagile Alpha-1 antitrypsin deficiency 338 Hemochromatosis Wilson disease
				clinical approach to a patient with a Storage disorders (MetabERN)		Fabry's disease: Gaucher's disease: Hunter's & Hurler's syndrome: Multiple carboxylase deficiency Niemann-pick lipidosis: Sanfilippo
		4	Chapter 24-3. Kidney disorders (ERKNet)	Polycystic renal disorders		PKD AD, PKD AR
				Functional disorder		Alport syndrome 340 Cystinosis Nail-patella
	S25. Genetics of Endocrine disorders					
		12	Chapter 25-1. Endocrine disorders (ENDO)	clinical approach to a patient with a Thyroid disorder		Primary congenital hypothyroidism (excluding secondary, transient, or other)

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				clinical approach to a patient with an Adrenal disorders clinical approach to a patient with Overgrowth clinical approach to a patient with a Short stature, proportionate clinical approach to a patient with Diabetes clinical approach to a patient with an anomaly of Lipid metabolism clinical approach to a patient with Obesity clinical approach to a patient with Lipodystrophy clinical approach to a patient with DSD		Congenital adrenal hyperplasia (CAH) 370 Dyschondrosteosis 98. SHOX, Noonan & CFC syndrome HNFIB-MODY 136. Familial hypercholesterolemia: Hyperlipidaemia 458 Prader-Willi Syndrome 144. Leptin deficiency Seip Androgen insensitivity syndrome (AIS) 344 Campomelic Dysplasia 126. SOX9, Persistent Millerian Duct Syndrome 51. AMH/MIS and Its Receptors:
	S26. Genetics of Skin disorders					
		10	Chapter 26-1. Skin disorders (SKIN)	clinical approach to a patient with alchtyosis clinical approach to a patient with an Ectodermal dysplasias clinical approach to a patient with an Epidermolysis bullosa clinical approach to a patient with a Pigmentary disorders & albinism clinical approach to a patient with a Disorders with photosensitivity		Biotinidase deficiency (including partial) Ichtyosis Anhidrotic ectodermal dysmplasia, XLR Ectodermal Dysplasia. Ectrodactyly, and Cleft Lip and/or Palate (EEC) 174. TP63 Incontinentia pigmenti Epidermolysis Albinism Progeria Syndrome Hutchinson-Gilford 171. Xeroderma pigmentosum & Cockayne
	S27. Genetics of Eye and vision disorders					
		10	Chapter 27-1. Eye and vision disorders (EYE)	clinical approach to a patient with a Anterior segment anomalies clinical approach to a patient with a Microphthalmos & coloboma		

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				clinical approach to a patient with Cataract		Galactosemia
				clinical approach to a patient with a Retinal disorders		Retinitis pigmentosa (RP) Bardet-Biedl)
				clinical approach to a patient with Optic atrophy		Leber optic atrophy
				clinical approach to a patient with a Color vision disorder		Daltonism
	S28. Genetics of Ear and hearing disorders					
		2	Chapter 28-1. Ear and hearing disorders (Cranio)	clinical approach to a patient with a Deafness		Usher Waardenburg
	S29. Genetics of Acute metabolic syndromes in neonates and children					
		6	Chapter 29-1. Acute metabolic syndromes in neonates and children (MetabERN)	clinical approach to a patient with a Hypoglycemia		Leigh encephalopathy 474 Medium-chain acyl-CoA dehydrogenase deficiency Very long-chain acyl-CoA dehydrogenase deficiency
				clinical approach to a patient with a Jaundice & liver problems		Citrullinemia type I Trifunctional protein deficiency
				clinical approach to a patient with a Metabolic acidosis		Argininosuccinic acidemia Glutaric acidemia type I Isovaleric acidemia Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency Maple syrup Urine disease: Methylmalonic acidemia (CblA,B, mutase deficiency) Propionic acidemia
	S30. Genetics of Skeletal disorders					
		8	Chapter 30-1. Skeletal disorders (BOND)	clinical approach to a patient with an Abnormal bone density		McCune-Albright/Fibrous Dysplasia 179. GNAS and Osteogenesis imperfecta Osteopetrosis Proteus Klippel-Trenaunay
				clinical approach to a patient with an Asymmetric growth		
				clinical approach to a patient with a Hyperlaxity - scoliosis		Ehlers Danlos syndromes (EDS) 406 Homocystinuria: Marfan & Loeys-Dietz Syndrome 53. TGFBR !/2 and Homocystinuria:

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				clinical approach to a patient with a Chondrodysplasia		Achondroplasia & hypochondroplasia Mukiple Synostosis (SYN 1), Tarsal-Carpal Coalition, and Isolated Stapes Ankylosis 47. NOG and Chondrodysplasia punctata Stickler syndrome
	S31. Psychiatric disorders					
		3	Chapter 31-1. Psychiatric disorders		Schizophrenia	
	S32. Genetics of Reproductive disorders					
		3	Chapter 32-1. Reproductive disorders	Recurrent miscarriages		Recurrent miscarriages
				Infertility		Klinefelter, Turner
Part 5. GENETICS IN MEDICINE : oncogenetics						
	S33. Genetic alterations in cancers					
		12	Chapter 33-1. Genetic alterations in cancers (GENTURIS)		Genetic bases of carcinogenesis Physiological functions of oncogenes and tumor suppressor genes MicroRNAs Epigenetic changes and tumors Driver mutations and passenger variants Expression profile and tumor signature	
	S34.Oncogenetics and the genetic predisposition to cancer					
		3	Chapter 34-1. Genetic predispositions to common cancers: breast cancer			Breast Cancer BRCA1, BRCA2, PALB2, RAD51
		3	Chapter 34-2. Genetic predispositions to common cancers: colon cancer and Lynch syndrome			GLynch MLH1, MSH2, MSH6, PMS2, EPCAM Familial adenomatous polyposis (FAP) and adenomatous polyposis due to non-APC MUTYH, POLE, POLD1, NTHL1

Part	Sections	# of pages	Chapters	COMPULSORY subchapters (perhaps one author per subchapter)	SUGGESTED topics/subchapters to cover Some may become subchapters with co-author	Links to clinical vignettes (may be > 1)
		3	**Chapter 34-3. Rare genetic predispositions to cancers**	**(may be subdivided in subgroups)**	General overview of rare predispositon to cancers: many disorders in vignettes (use tables and link to vignettes)	**Birt-Hogg-Dubé FLCN** **AXIN2, Tooth Agenesis, and Colorectal Cancer 37.** **Cowden (PTEN hamartoma tumour syndrome (PHTS)) 564 , PIK3CA, AKT1** **Diffuse Gastric Cancer (HDGC) CDH1** **Familial pituitary cancers AIP** **Fanconi Anemia 172.** **Gorlin Basal Cell Nevus Syndrome 31.** **Hereditary papillary carcinoma FH, MET** **Juvenile polyposis BMPR1A, SMAD4 (JPS) 578** **Li-fraumeni TP53, CHEK2 (LFS) 584** **Melanoma malignant, familial CDKN2A, MITF, BAP1, POT1, CDK4** **MEN 1** **MEN2 69. RET** **Neurofibromatosis type 1 (NF1) 506** **Neurofibromatosis type 2 (NF2) 596** **Ovarian cancer** **Paraganglioma-pheochromocytome syndrome SDH, TMEM127, MAX, EPAS1** **Peutz-Jeghers 77. STK11 and**
Part 6. GENETICS IN MEDICINE : Clinical vignettes						
	S35. Vignettes					
			Chapter 35(see specific xls file)		General list Cancer-predisposing list	
Part 7. Personalized medicine						
	S36. Personalized genomic medicine					
		3	**Chapter 36-1. Pharmacogenetics**			
		3	**Chapter 36-2. Theranostics**			
		4	**Chapter 36-3. Personalized genomic medicine**		Personalized genomic medicine Genetic profiling (recreational genetics)	
Part 8. Screening for genetic disease						
	S37. Postnatal genetic screening					
		2	**Chapter 37-1. Family screening**			
		2	**Chapter 37-2. Post-natal population screening**			
		4	**Chapter 37-3. Prenatal screening**			

Part	Sections	# of pages	Chapters	COMPULSORY subchapters (perhaps one author per subchapter)	SUGGESTED topics/subchapters to cover Some may become subchapters with co-author	Links to clinical vignettes (may be > 1)
	S38. Prenatal genetic diagnosis					
		5	**Chapter 38-1. Prenatal diagnosis by imaging**			
		10	**Chapter 38-2. Invasive prenatal diagnosis**			
		3	**Chapter 38-3. Non-invasive prenatal diagnosis**			
Part 9. Treatment of genetic diseases						
	S39. Indirect treatment					
		2	**Chapter 39-1. Non-specific phenotype-targeting treatments**			
		2	**Chapter 39-2. Indirect pharmacological therapies**			
		2	**Chapter 39-3. Therapeutics targeting abnormal protein**		Substitutive enzymotherapies	
		2	**Chapter 39-4. Action on gene expression**			
		2	**Chapter 39-5. Cell therapy**		synthetic lethality (PARPP inhibitors in BRCA-deficient cancer)	
	S40. Gene therapy and gene editing					
		4	**Chapter 40-1. Gene therapy**			
		4	**Chapter 40-2. Gene editing**			
		2	**Chapter 40-3. Therapeutic trials in genetic diseases**			
Part 10. Genetics on the web						
	S41. Genetics on the web					
		2	**Chapter 41-1. Normal genome (UCSC, Ensembl…)**			
		2	**Chapter 41-2. Proteins databases**			
		2	**Chapter 41-3. Variation databases (ClinVar, Gnomad, DECIPHER…)**			
		2	**Chapter 41-4. Integrative websites (Genecard, Varsome…)**			
		2	**Chapter 41-5. Clinical resources (OMIM, Orphanet, Genereviews…)**			
AUTO-EVALUATION						