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Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
Part 1. HUMAN GENETICS						
	S1. Introduction to Human Genetics					
		5	**1-1**	**Chapter 1-1. Introduction: genetics and medicine**		
						Medical genetics *Genetic diseases* *The heterogeneity of genetic diseases* *Rare diseases*
	S2. Genomics					
		10	**2-1**	**Chapter 2-1. Basics of nucleic acids biology**		
						DNA biochemistry *The genetic code* *Organization of human genes* *Coding and functional, non-non-coding RNAs* *Chromatin* *Organization of human genome*
		10	**2-2**	**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	**2-3**	**Chapter 2-3. Regulation of gene translation**		
						Mechanism of translation *Post-translational modifications*
		10	**2-4**	**Chapter 2-4. Mitosis and meiosis**		
						DNA replication and chromosome formation *Cell cycle and its regulation* *Mitosis* *Méiosis*
		5	**2-5**	**Chapter 2-5. Transmission of the genome**		
						Transmission of the genome & gametogenesis

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Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
	S3. Variants et mutations					
		15	**3-1**	**Chapter 3-1. Qualitative and quantitative variations of the genome**		
						Variations affecting the sequence of a gene
						Dynamic variations
						Structural genomic rearrangements, balanced
						Structural genomic rearrangements unimbalanced
						Anomaly in the number of chromosomes
						Mosaicism
						UPD
		10	**3-2**	**Chapter 3-2. Functional consequences of mutations on proteins (selected examples)**		
						Enzymes
						Receptors
						Channels
						Signal transduction
						Structural proteins
		5	**3-3**	**Chapter 3-3. Genotype/phenotype relationships**		
						Basis of dominant inheritance
						Basis of recessive inheritance
		5	**3-4**	**Chapter 3-4. Mechanism for the emergence of mutations**		
						Mechanism of mutation & triggers
		5	**3-5**	**Chapter 3-5. Descriptive nomenclature of gene and genomic anomalies**		
						Descriptive nomenclature of gene anomalies (HGVS)
						Descriptive nomenclature of cytogenomic anomalies (ISCN)
		10	**3-6**	**Chapter 3-6. Formal genetics: heredity modes**		
						Autosomal dominant heredity
						Autosomal recessive heredity
						X-linked heredity
						Pseudoautosomal heredity

Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
						Chromosomal inheritance *Mitochondrial inheritance* *Non-mendelian inheritance* *Mosaicism* *Uniparental disomy*
	S4. Multifactorial/genetic predisposition to common diseases					
		15	**4-1**	**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	**5-1**	**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	**6-1**	**Chapter 6-1. Classical technologies**		
						Historical aspects: positional cloning of disease-causing genes *Detection of short sequence variants* *PCR*

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Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
						Sanger sequencing *Expansion detection* *Methylation detection* *Quantitative dosages* *Classical cytogenetics and FISH*
		10	**6-2**	**Chapter 6-2. Genomics technologies**		
						Molecular cytogenetics: CGH and SNP arrays *NGS technologies (short reads)* *NGS technologies (long reads)* *Detecting structural rearrangements (Nano…)*
		3	**6-3**	**Chapter 6-3. Transcriptomics technologies**		
						RNA seq *Gene expression profiling*
		5	**6-4**	**Chapter 6-4. Epigenomics technologies**		
						Investigation of chromatine architecture *ATAC-Seq* *Conformation capture technologies*
		2	**6-5**	**Chapter 6-5. Proteomics and metabolomics technologies**		
						Mass spectrometry
		5	**6-6**	**Chapter 6-6. Establish the pathogenicity of a variant**		
						Evaluating pathogenicity *ACMG classification*
	S7. Clinical genetics and genetic counseling					
		5	**7-1**	**Chapter 7-1. Basics of statistics applied to genetics**		
						Risk calculation *Bayes*
		10	**7-2**	**Chapter 7-2. Genetic counseling**		
						Pedigree drawing *Genetic counseling in Mendelian disorders* *Genetic counseling in genomic disorders*

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Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
						Genetic counseling in common disorders *Genetic clinics : practical aspects*
		5	**7-3**	**Chapter 7-3. Carrier testing in families**		
						Carrier testing in families
		5	**7-4**	**Chapter 7-4. Presymptomatic diagnosis**		
						Presymptomatic:predictive diagnosis
		3	**7-5**	**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	**7-6**	**Chapter 7-6. The management of rare diseases in EU and the ERNs**		
						The management of rare diseases in EU and the ERNs
	S8. The genetics of developmental anomalies					
		5	**7-7**	**Chapter 8-1. Developmental genetics**		
						Genetic control of development *Epigenetic control of development (examples by organ are in the pathway section)*
				Chapter 8-2. Genetic of developmental pathways (may be discussed with ERNs if embryologists)		
		2	**8-2-1**		**DP1: Early embryonic development and Left-Right Asymmetry**	
		2	**8-2-2**		**DP2: Neural Crest Formation and Craniofacial Development**	
		2	**8-2-3**		**DP3: Development of the Nervous System: neural tube**	
		2	**8-2-4**		**DP4: Development of the Nervous System: neuronogenesis**	
		2	**8-2-5**		**DP5. Development of the Eye**	
		2	**8-2-6**		**DP6. Development of the Ear**	
		2	**8-2-7**		**DP7. Muscle and Somite Development**	
		2	**8-2-8**		**DP8. Bone and Cartilage**	
		2	**8-2-9**		**DP9. Development of the limbs**	
		2	**8-2-10**		**DP10. Cardiogenesis**	
		2	**8-2-11**		**DP11. Kidney**	
		2	**8-2-12**		**DP12. Development of the Endodermal Derivatives: Lung, Liver, Pancreas, and Gut**	
		2	**8-2-13**		**DP13. Development of skin and Epidermal Appendages: Teeth and Hair**	

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		2	8-2-14		DP14. Control of sex determination	
Part 3. GENETICS IN MEDICINE : etiopathogenic approach						
	S9. Genetic basis of metabolic processes					
				Chapter 9-1. Metabolic disorders (inborn errors)		
		2	9-1-1		A1. amino acid metabolism & urea cycle	
		2	9-1-2		A2. carbohydrate metabolism	
		2	9-1-3		A3. fatty acid, carnitine and ketone body	
		2	9-1-4		A4. Mitochondria: energy metabolism	
		2	9-1-5		A5. lipid metabolism	
		2	9-1-6		A6. lipoprotein metabolism	
		2	9-1-7		A7. Complex molecule degradation –lysosomal disorders	
		2	9-1-8		A8. Peroxisomes	
		2	9-1-9		A9. purine, pyrimidine and nucleotide metabolism	
		2	9-1-10		A10. Cofactors ands vitamins	
		2	9-1-11		A11. Porphirins	
		2	9-1-12		A12. Steroids	
		2	9-1-13		A13. Glycosylation (O and N)	
		2	9-1-14		A14. Metal	
	S10. Genetic basis of Intracellular signalling					
				Chapter 10-1. Signaling pathways (15)		
		2	10-1-1		Cell receptors overview	
		2	10-1-2		C1. B: SHH Signaling Pathway	
		2	10-1-3		C2. C: Wnt Signaling Pathway	
		2	10-1-4		C3. A: TGF-b Signaling Pathway	
		2	10-1-5		C4. C: FGF Signaling Pathway	
		2	10-1-6		C5. D: GDNF Signaling Pathway	
		2	10-1-7		C6. G:RTK/RAS/MAP & PI3K/AKT pathways	
		2	10-1-8		C7. I: EPHRIN Signaling Pathway	
		2	10-1-9		C8. TNF Signaling pathway	
		2	10-1-10		C9:NOG pathway	
		2	10-1-11		C10: MYC pathway	
		2	10-1-12		C11: Notch pathway	
		2	10-1-13		C12: p53 pathway	

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		2	10-1-14		C13: DNA repair	
		2	10-1-15		F4. D: GUANINE NUCLEOTIDE-BINDING PROTEINS	
				Chapter 10-2. Transcription factors		
		2	10-2-1		D1. A: The Homeobox Gene Family	
		2	10-2-2		D2. B: The Paired Box (PAX) Gene Family	
		2	10-2-3		D3. C: The Forkhead Gene Family	
		2	10-2-4		D4. D: The T-Box Gene Family	
		2	10-2-5		D5. E: THE SOX gene family	
		2	10-2-6		D6: other TF	
				Chapter 10-3. Regulation of chromatin function		
		2	10-3-1		E1. Histone/DNA méthylation regulation	
		2	10-3-2		E2. Cohesines	
				Chapter 10-4. Regulation of protein synthesis		
		2	10-4-1		F1+F7. A: RNA processing (localisation, spliceosome)	
		2	10-4-2		F2. B: Post-translational control & ubiquitinisation	
	S11. Genetic basis of cell dynamics					
				Chapter 11-1. Cellular dynamics		
		2	11-1-1		F3. C:Cell cycle and apoptosis	
		2	11-1-2		F5. E: Cytoskeletton	
		2	11-1-3		F6. F: Vesicle-mediated trafficking	
	S12. Genetic basis of Extracellular matrix					
		2	12-1	Chapter 12-1. Extracellular matrix		
					G1. Collagen & fibrillin	
	S13. Genetic basis of Junctions, transporters and channels					
		8	13-1	Chapter 13-1. Junctions, transporters and channels		
					H1. GAP junctions	
					H2. Channels	
					H3. Membrane transporter	
					H4. Membrane proteins	
	S14. Genetic basis of Ciliary Functions: Genesis, Transport, and Reabsorbtion					
		3	14-1	Chapter 14-1. Ciliary Functions: Genesis, Transport, and Reabsorbtion		
					B1. Primary cilium and its function	
	S15. Genetic basis of red cell metabolism					

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		4	**15-1**	**Chapter 15-1. Red cell metabolism**		
						I1. Heme synthesis *I2. Hemoglobin synthesis*
	S16. Genetic basis of Hemostasis					
		3	**16-1**	**Chapter 16-1. Hemostasis**		
						J1. Coagulation cascade *J2. Platelet function*
	S17. Genetic basis of Immune system					
		3	**17-1**	**Chapter 17-1. Immune system**		
						K1. Ig and T cell receptors
Part 4. GENETICS IN MEDICINE : clinically oriented approach						
	S18. Dysmorphology and congenital malformations					
		5	**18-1**	**Chapter 18-1. Congenital malformations**		
						Congenital malformations: epidemiology & classification
		5	**18-2**	**Chapter 18-2. Dysmorphology**		
						Basics of dysmorphology *Examination of a child with developmental anomalies*
				Chapter 18-3. Craniofacial anomalies		
		2	**18-3-1**		**Craniosynostosis**	
		2	**18-3-2**		**Cleft & Robin sequence**	
		2	**18-3-3**		**Ear anomalies**	
		2	**18-3-4**		**Oculopalpebral anomalies**	
				Chapter 18-4. Selected malformations		
		2	**18-4-1**		**Limb malformation**	
		2	**18-4-2**		**Congenital heart disorders**	
		2	**18-4-3**		**Neural tube defects**	
		1			*Multiple congenital anomalies (selected examples as vignettes)*	
	S19. Environmentallly-induced birth defects					
		2	**19-1**	**Chapter 19-1. Chemical exposure**		
						Teratogenic exposure
		2	**19-2**	**Chapter 19-2. Infections**		

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		4	**19-3**	**Chapter 19-3. Irradiation**		
						Effect of irradiations
	S20. Genetic of neurodevelopment, including ID and ASD					
		10	**20-1**	**Chapter 20-1. Genetic and neurodevelopment, including ID and ASD**		
						Medical definition of intelligence *Intellectual disability and NDD: evaluation* *Autism spectrum disorder*
						Etiologies and epidemiology of ID and ASD
						Pathophysiology *Diagnostic strategies* *Medical care*
		1				*Syndromal ID syndromes (selected examples)*
	S21. Genetics of Central and peripheral nervous system					
		5	**21-1**	**Chapter 21-1. Brain malformations (see with EPICARE)**		
						Holoprosencephaly *Microcephaly and lissencephaly* *Cerebellum and brainstem* *Hydrocephalus & corpus callosum*
		15	**21-2**	**Chapter 21-2. Neurological disorders (see with RND and EPICARE)**		
						Epilepsy *Hypotonia* *Spastic paraplegia* *Ataxia* *Neurodegenerative disorder in children* *Neurodegenerative disorders in adults* *Neurovascular diseases* *Neuropathies*
		5	**21-3**	**Chapter 21-3. Neuromuscular disorders (see with ERN NMD)**		
						Muscle disorders *Muscular dystrophies* *Arthrogryposis multiplex*

Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
	S22. Genetics of Heart and vessels disorders					
		3	**22-1**	**Chapter 22-1. Heart disorders (See with HEART)**		
						Cardiomyopathies
						Long QT and other ryhthmic disorders
		3	**22-2**	**Chapter 22-2. Vessels disorders (se with VASCERN)**		
						Vascular disorders
	S23. Genetics of Immuno-hematological disorders					
		3	**23-1**	**Chapter 23-1. Coagulation disorders (See with EuroBloodNet)**		
						Hemolysis
						Coagulation defect
		3	**23-2**	**Chapter 23-2. Hemoglobin disorders (See with EuroBloodNet)**		
						Hemoglobinopathies
		3	**23-3**	**Chapter 23-3. Immune disorders (see with ERN RITA)**		
						B cell immunodeficiencies
						T cell Immunodeficiencies
						SCID
	S24. Genetics of Visceral disorders					
		2	**24-1**	**Chapter 24-1. Pulmonary disorders (see with LUNG ERN)**		
						Cystic fibrosis
						Surfactant
		4	**24-2**	**Chapter 24-2. Liver, pancreas and digestive tract disorders see with LIVER partim)**		
						digestive tube
						Cirrhosis
						Storage disorders
		4	**24-3**	**Chapter 24-3. Kidney disorders (see with ERKNet)**		
						Polycystic renal disorders
						Functional disorder
	S25. Genetics of Endocrine disorders					
		12	**25-1**	**Chapter 25-1. Endocrine disorders (see with ENDO-ERN)**		
						Thyroid disorders
						Adrenal disorders
						Short stature, proportionate
						Diabetes

Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
						Lipid metabolism *Obesity* *Lipodystrophy* *DSD*
	S26. Genetics of Skin disorders					
		10	**26-1**	**Chapter 26-1. Skin disorders (See with SKIN)**		
						Ichtyosis *Ectodermal dysplasias* *Epidermolysis bullosa* *Pigmentary disorders & albinism* *Disorders with photosensitivity*
	S27. Genetics of Eye and vision disorders					
		10	**27-1**	**Chapter 27-1. Eye and vision disorders (see with EYE)**		
						Anterior segment anomalies *Microphthalmos & coloboma* *Cataracts* *Retinal disorders* *Optic atrophy* *Color vision*
	S28. Genetics of Ear and hearing disorders					
		2	**28-1**	**Chapter 28-1. Ear and hearing disorders**		
						Deafness
	S29. Genetics of Acute metabolic syndromes in neonates and children					
		6	**29-1**	**Chapter 29-1. Acute metabolic syndromes in neonates and children (see with MetabERN)**		
						Hypoglycemia *Jaundice & liver problems* *Metabolic acidosis*
	S30. Genetics of Skeletal disorders					
		8	**30-1**	**Chapter 30-1. Skeletal disorders (see with ERN BOND)**		
						Abnormal bone density

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						Asymmetric growth
						Hyperlaxity - scoliosis
						Chondrodysplasia
	S31. Psychiatric disorders					
		3	**31-1**	**Chapter 31-1. Psychiatric disorders**		
						Schizophrenia
	S32. Genetics of Reproductive disorders					
		3	**32-1**	**Chapter 32-1. Reproductive disorders**		
						Recurrent miscarriages
						Infertility
Part 5. GENETICS IN MEDICINE : oncogenetics						
	S33. Genetic alterations in cancers					
		12	**33-1**	**Chapter 33-1. Genetic alterations in cancers (see with 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	**34-1**	**Chapter 34-1. Genetic predispositions to common cancers: breast cancer**		
		3	**34-2**	**Chapter 34-2. Genetic predispositions to common cancers: colon cancer and Lynch syndrome**		
		3	**34-3**	**Chapter 34-3. Rare genetic predispositions to cancers**		
Part 6. GENETICS IN MEDICINE : Clinical vignettes						
	S35. Vignettes					
				Will be discussed with Orphanet		*General list*
						Cancer-predisposing list
Part 7. Personalized medicine						
	S36. Personalized genomic medicine					
		3	**36-1**	**Chapter 36-1. Pharmacogenetics**		
						Pharmacogenetics
		3	**36-2**	**Chapter 36-2. Theranostics**		

Part	Section	# of pages	Chapter ID	Chapters One author per chapter (Normally)	Mandatory subchapters (Perhaps one author per subchapter)	Suggested subchapters (TBC by the author)
						Theranostics
		4	**36-3**	**Chapter 36-3. Personalized genomic medicine**		
						Personalized genomic medicine
						Genetic profiling (recreational genetics)
Part 8. Screening for genetic disease						
	S37. Postnatal genetic screening					
		2	**37-1**	**Chapter 37-1. Family screening**		
						Family screening
		2	**37-2**	**Chapter 37-2. Post-natal population screening**		
						Population screening
		4	**37-3**	**Chapter 37-3. Prenatal screening**		
						Prenatal screening
	S38. Prenatal genetic diagnosis					
		5	**38-1**	**Chapter 38-1. Prenatal diagnosis by imaging**		
						Fetal imaging
		10	**38-2**	**Chapter 38-2. Invasive prenatal diagnosis**		
						Invasive diagnosis
		3	**38-3**	**Chapter 38-3. Non-invasive prenatal diagnosis**		
						NIPT/NIPD
Part 9. Treatment of genetic diseases						
	S39. Indirect treatment					
		2	**39-1**	**Chapter 39-1. Non-specific phenotype-targeting treatments**		
						Non specific
		2	**39-2**	**Chapter 39-2. Indirect pharmacological therapies**		
						Pharmaco
		2	**39-3**	**Chapter 39-3. Therapeutics targeting abnormal protein**		
						Abnormal protein
		2	**39-4**	**Chapter 39-4. Action on gene expression**		
						Gene expression
		2	**39-5**	**Chapter 39-5. Cell therapy**		
						Cell therapy
	S40. Gene therapy and gene editing					
		4	**40-1**	**Chapter 40-1. Gene therapy**		

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						Gene thrapy
		4	**40-2**	**Chapter 40-2. Gene editing an CRSPR-Cas9**		
						Gene editing
		2	**40,3**	**Chapter 40-3. Therapeutic trials in genetc diseases**		
						Therapeutic trials in genetc diseases
Part 10. Genetics on the web						
	S41. Genetics on the web					
						Internet
		2	**41-1**	**Chapter 41-1. Normal genome (UCSC, Ensembl…)**		
						Genome
		2	**41-2**	**Chapter 41-2. Proteins databases**		
						Proteins
		2	**41-3**	**Chapter 41-3. Variation databases (ClinVar, Gnomad, DECIPHER…)**		
						Variants
		2	**41-4**	**Chapter 41-4. Integrative websites (Genecard, Varsome…)**		
						Integrative *Med encyclopedia*
		2	**41-5**	**Chapter 41-5. Clinical resources (OMIM, Orphanet, Genereviews…)**		
						Medical
AUTO-EVALUATION						