Important Information in tables

TABLE 7-2 Glycogen Storage Disorders

TYPE	DEFECT	MAJOR AFFECTED TISSUES
Ia (Von Gierke)	Glucose-6-phosphatase	Liver, kidney, intestine
Ib	Microsomal glucose-6-phosphate transport	Liver, kidney, intestine, neutrophils
II (Pompe)	Lysosomal acid β-glucosidase	Muscle, heart
IIIa (Cori)	Glycogen debranching enzyme	Liver, muscle
IIIb	Glycogen debranching enzyme	Liver
IV (Anderson)	Branching enzyme	Liver, muscle
V (McArdle)	Muscle phosphorylase	Muscle
VI (Hers)	Liver phosphorylase	Liver
VII (Tarui)	Muscle phosphofructokinase	Muscle

TABLE 7-3 Mucopolysaccharidoses*

NAME	MUTANT ENZYME	CLINICAL FEATURES
Hurler–Scheie (MPS-I)	α-L-Iduronidase	Coarse face, hepatosplenomegaly, corneal clouding, dysostosis multiplex,† intellectual disability
Hunter (MPS-II)	Iduronate sulfatase	Coarse face, hepatosplenomegaly, dysostosis multiplex, intellectual disability, behavioral problems
Sanfilippo A MPS-IIIA	Heparan-*N*-sulfamidase	Behavioral problems, dysostosis multiplex, intellectual disability
Sanfilippo B MPS-IIIB	α-*N*-Acetylglucosaminidase	Behavioral problems, dysostosis multiplex, intellectual disability
Sanfilippo C MPS-IIIC	Acetyl-CoA:α-glucosaminide *N*-acetyltransferase	Behavioral problems, dysostosis multiplex, intellectual disability
Sanfilippo D MPS-IIID	*N*-Acetylglucosamine-6-sulfatase	Behavioral problems, dysostosis multiplex, intellectual disability
Morquio A MPS-IVA	*N*-Acetylglucosamine-6-sulfatase	Short stature, bony dysplasia, hearing loss
Morquio B MPS-IVB	β-Galactosidase	Short stature, bony dysplasia, hearing loss
Maroteaux–Lamy MPS-VI	Aryl sulfatase B	Short stature, corneal clouding, cardiac valvular disease, dysostosis multiplex
Sly MPS-VII	β-Glucuronidase	Coarse face, hepatosplenomegaly, corneal clouding, dysostosis multiplex

*Hunter syndrome is an X-linked recessive disorder; the remaining MPS disorders are autosomal recessive.

†Dysostosis multiplex is a distinctive pattern of changes in bone, including a thickened skull, anterior thickening of the ribs, vertebral abnormalities, and shortened and thickened long bones.

TABLE 7-4 Lysosomal Storage Disorders*

NAME	MUTANT ENZYME	CLINICAL FEATURES
Tay–Sachs	β-Hexosaminidase A	Hypotonia, spasticity, seizures, blindness
Gaucher (type 1; nonneuropathic)	β-Glucosidase	Splenomegaly, hepatomegaly, bone marrow infiltration, brain usually spared
Niemann–Pick, type 1A	Sphingomyelinase	Hepatomegaly, corneal opacities, brain deterioration
Fabry	α-Galactosidase	Paresthesia of the hands and feet, corneal dystrophy, hypertension, renal failure, cardiomyopathy
G_{M1} gangliosidosis (infantile)	β-Galactosidase	Organomegaly, dysostosis multiplex,† cardiac failure
Krabbe	galactosylceramidase	Hypertonicity, blindness, deafness, seizures, (galactosylceramide-specific) atrophy of the brain
Metachromatic leukodystrophy	Aryl sulfatase A	Ataxia, weakness, blindness, brain atrophy (late-infantile)
Sandhoff	β-Hexosaminidase (total)	Optic atrophy, spasticity, seizures
Schindler	α-*N*-Acetylgalactosaminidase	Seizures, optic atrophy, retardation
Multiple sulfatase deficiency	Aryl sulfatase A, B, C	Retardation, coarse facial features, weakness, hepatosplenomegaly, dysostosis multiplex

*Of the lysosomal storage disorders included in this table, Fabry syndrome is X-linked recessive and the remainder are autosomal recessive.
†Dysostosis multiplex is a distinctive pattern of changes in bone, including a thickened skull, anterior thickening of the ribs, vertebral abnormalities, and shortened and thickened long bones.

Tables 9-1, 9-2 and 9-3: Just the related info that is mentioned in slides text

TABLE 9-2 Examples of Major Histocompatibility Complex and Disease Associations

DISEASE	MHC (HLA) ASSOCIATED LOCUS (LOCI)*	APPROXIMATE RELATIVE RISK†
Type 1 diabetes	DQB1, DQA1	10
Ankylosing spondylitis	B27	90
Narcolepsy	DR2 and DQA1, DQB1	>100
Celiac disease	DQA1, DQB1	10
Rheumatoid arthritis	DRB1, DQA1	5
Myasthenia gravis	C, DR3, DR7	2.5
Multiple sclerosis	DRB1	4
Systemic lupus erythematosus	DRB1	6
Hemochromatosis	A3	20
Malaria	B53	0.6059
Graves disease	DR3	5
Psoriasis	C	13
Abacavir (anti-HIV drug) hypersensitivity	B57	≈1000

*For simplicity, specific alleles are not shown here. For example, the *HLA-B57* allele associated with abacavir sensitivity is labeled *HLA-B*57:01,* and the allele associated with psoriasis is *HLA-C*06:02.*

†Relative risk can be interpreted loosely as the odds that a person who has a risk factor (in this case, an MHC antigen) will develop the disease, compared with a person who lacks the risk factor. Thus, a relative risk of 4 for *DRB1* and multiple sclerosis means that persons with a specific *DRB1* allele are four times more likely to develop multiple sclerosis than are those without it. A relative risk <1 (as seen for malaria and *B53*) indicates that the factor is protective against the disease. These relative risks can vary among different human population groups.

Tables 10-2 : Just the related info that is mentioned in slides text

TABLE 10-1 Animal Models of Human Development

ORGANISM	GENERATION TIME*	ADVANTAGES	DISADVANTAGES
Caenorhabditis elegans (roundworm)	9 days	Fate of every cell known Genome well characterized Easy to breed and maintain	Alternative body plan compared to vertebrates Tissues cannot be cultured
Drosophila melanogaster (fruit fly)	10 days	Easy to breed Large populations Vast database of mutants Feasible and affordable to do large screens	Alternative body plan compared to vertebrates Must be stored live; cannot be frozen Pathology often different compared to humans
Danio rerio (zebrafish)	3 months	Transparent embryo Easy to maintain Large populations Feasible and affordable to do large screens	Targeted gene modification difficult
Xenopus laevis (frog)	12 months	Transparent embryo is large and easy to manipulate	Tetraploid genome makes genetic experiments difficult
Gallus gallus (chicken)	5 months	Easy to observe and manipulate embryo	Genetic experiments difficult
Mus musculus (mouse)	2 months	Pathology similar to humans Excellent tools for phenotypic characterization Targeted gene modification straightforward Fully annotated genome available	Relatively expensive to maintain Manipulation of embryo is challenging
Papio hamadryas (baboon)	60 months	Pathology and physiology similar to that of humans	Very expensive to maintain Small populations Long generation time Strong ethical concerns with use of primates

*Generation time is defined as the age at which the organism is first capable of reproduction.

Tables 11-1 and 11-3: Just the genes that are mentioned in the slides text

TABLE 11-2 Comparison of Key Features of Tumor Suppressor Genes and Oncogenes

FEATURE	TUMOR SUPPRESSOR GENES	ONCOGENES
Function of normal version	Regulates cell growth and proliferation; some can induce apoptosis	Promotes cell growth and proliferation
Mutation (at cell level)	Recessive (both copies of gene inactivated)	Dominant (only one copy of gene mutated)
Effect of mutation	Loss of function	Gain of function
Germline mutations resulting in inherited cancer syndromes	Seen in most tumor suppressor genes	Seen in only a few oncogenes

Tables 12-4 and 12-5: Just the related info that is mentioned in slides text

TABLE 12-2 Recurrence Risks for First-, Second-, and Third-Degree Relatives of Probands

DISEASE	PREVALENCE IN GENERAL POPULATION	DEGREE OF RELATION		
		FIRST DEGREE	SECOND DEGREE	THIRD DEGREE
Cleft lip/palate	0.001	0.04	0.007	0.003
Club foot	0.001	0.025	0.005	0.002
Congenital hip dislocation	0.002	0.005	0.006	0.004

TABLE 14-1 Examples of Effects of Gene Polymorphisms on Drug Response

GENE	ENZYME/TARGET	DRUG	CLINICAL RESPONSE
CYP2D6	Cytochrome P4502D6	Codeine	Persons homozygous for an inactivating mutation do not metabolize codeine to morphine and thus experience no analgesic effect
CYP2C9	Cytochrome P4502C9	Warfarin	Persons heterozygous for a polymorphism need a lower dose of warfarin to maintain anticoagulation
VKORC1	Vitamin K epoxide reductase	Warfarin	Persons heterozygous for a polymorphism need a lower dose of warfarin complex, subunit 1, to maintain anticoagulation
NAT2	*N*-Acetyl transferase 2	Isoniazid	Persons homozygous for slow-acetylation polymorphisms are more susceptible to isoniazid toxicity
TPMT	Thiopurine S-methyltransferase	Azathioprine	Persons homozygous for an inactivating mutation develop severe toxicity if treated with standard doses of azathioprine
ADRB2	β-Adrenergic receptor	Albuterol	Persons homozygous for a polymorphism get worse with regular use of albuterol
KCNE2	Potassium channel, voltage-gated	Clarithromycin	Persons heterozygous for a polymorphism are more susceptible to life-threatening arrhythmias
SUR1	Sulfonylurea receptor 1	Sulfonylureas	Persons heterozygous for polymorphisms exhibit diminished sensitivity to sulfonylurea-stimulated insulin secretion
F5	Coagulation factor V (Leiden)	Oral contraceptives	Persons heterozygous for a polymorphism are at increased risk for venous thrombosis

TABLE 4-1 A Comparison of the Major Attributes of Autosomal Dominant and Autosomal Recessive Inheritance Patterns

ATTRIBUTE	AUTOSOMAL DOMINANT	AUTOSOMAL RECESSIVE
Usual recurrence risk	50%	25%
Transmission pattern	Vertical; disease phenotype seen in generation after generation	Disease phenotype may be seen in multiple siblings, but usually not in earlier generations
Sex ratio	Equal number of affected males and females (usually)	Equal number of affected males and females (usually)
Other	Father-to-son transmission of disease gene is possible	Consanguinity is sometimes seen, especially for rare recessive diseases

TABLE 5-2 Comparison of the Major Attributes of X-Linked Dominant and X-Linked Recessive Inheritance Patterns*

ATTRIBUTE	X-LINKED DOMINANT	X-LINKED RECESSIVE
Recurrence risk for heterozygous female × normal male mating	50% of sons affected; 50% of daughters affected	50% of sons affected; 50% of daughters heterozygous carriers
Recurrence risk for affected male × normal female mating	0% of sons affected; 100% of daughters affected	0% of sons affected; 100% of daughters heterozygous carriers
Transmission pattern	Vertical; disease phenotype seen in generation after generation	Skipped generations may be seen, representing transmission through carrier females
Sex ratio	Twice as many affected females as affected males (unless disease is lethal in males)	Much greater prevalence of affected males; affected homozygous females are rare
Other	Male-to-male transmission is not seen; expression is less severe in female heterozygotes than in affected males	Male-to-male transmission not seen; manifesting heterozygotes may be seen in females

*Compare with the inheritance patterns for autosomal diseases shown in Table 4-1.