Pattern of inheritance

Disease / Disorder	Inheritance Pattern	Genetic Concept Highlighted in Lecture	Key Point
Retinoblastoma	**Autosomal Dominant**	**Reduced penetrance**	~90% penetrance; some mutation carriers are unaffected.
Waardenburg Syndrome	**Autosomal Dominant**	**Reduced penetrance**	Syndromic deafness, heterochromia, white forelock; only ~20% are deaf.
Neurofibromatosis	**Autosomal Dominant**	**Variable expressivity**	Disease always present but severity varies greatly.
Myotonic Dystrophy	**Autosomal Dominant**	Variable expressivity + anticipation + trinucleotide repeat expansion	Severity and age of onset vary; worsens in successive generations.
Huntington Disease	**Autosomal Dominant**	Variable age of onset + trinucleotide repeat disease	Usually begins age 35–40; anticipation may occur.
Marfan Syndrome	**Autosomal Dominant**	**Pleiotropy**	Skeletal, cardiovascular, and ocular manifestations.
Cystic Fibrosis (CF)	**Autosomal Recessive**	Pleiotropy + Allelic heterogeneity	Many CFTR mutations can cause the same disease.
Duchenne Muscular Dystrophy (DMD)	**X-linked Recessive**	Allelic heterogeneity	Different mutations in same gene produce disease.
Becker Muscular Dystrophy (BMD)	**X-linked Recessive**	Allelic heterogeneity	Less severe dystrophinopathy than DMD.

Disease / Disorder	Inheritance Pattern	Genetic Concept Highlighted in Lecture	Key Point
Congenital Hearing Loss (Deafness)	Can be **AD or AR**	**Locus heterogeneity**	Many different genes can cause the same phenotype.
Autosomal Dominant Hearing Loss (PAX3)	**Autosomal Dominant**	Locus heterogeneity example	Mutation in PAX3 gene.
Autosomal Recessive Hearing Loss (GJB2)	**Autosomal Recessive**	Locus heterogeneity example	Mutation in GJB2 gene.
Male Precocious Puberty	**Autosomal Dominant**	**Sex-limited trait**	Gene inherited in both sexes but expressed only in males.
Hemochromatosis	**Autosomal Recessive**	**Sex-influenced trait**	More common/severe in males.
Hemophilia	**X-linked Recessive**	Sex-linked inheritance	Gene located on X chromosome.
Fragile X Syndrome (FMR1)	**X-linked dominant-like atypical inheritance**	Trinucleotide repeat expansion, anticipation, parent-of-origin effect	CGG expansion; full mutation >200 repeats.
Fragile X–Associated Tremor/Ataxia Syndrome (FXTAS)	Related to **FMR1 premutation** **In males**	Dynamic mutation consequence	Occurs in some premutation carriers.
Premature Ovarian Failure (POF)	Associated with **FMR1 premutation** **In Females**	Dynamic mutation consequence	Seen in some female premutation carriers.
Spinocerebellar Ataxia	Usually **Autosomal Dominant**	Trinucleotide repeat disease	Repeat expansion disorder.

Disease / Disorder	Inheritance Pattern	Genetic Concept Highlighted in Lecture	Key Point
Kennedy Disease	**X-linked Recessive**	Trinucleotide repeat disease	Repeat expansion disorder.
Joseph Disease (Machado–Joseph Disease)	**Autosomal Dominant**	Trinucleotide repeat disease	Repeat expansion disorder.
Friedreich Ataxia	**Autosomal Recessive**	Trinucleotide repeat disease	Repeat expansion disorder.
Prader–Willi Syndrome (PWS)	**Genomic imprinting disorder**	Paternal 15q11-q13 loss / maternal UPD	Obesity, hyperphagia, intellectual disability.
Angelman Syndrome (AS)	**Genomic imprinting disorder**	Maternal 15q11-q13 loss / paternal UPD / UBE3A mutation	Happy demeanor, ataxia, intellectual disability.
Mitochondrial Myopathy	**Mitochondrial (Maternal) inheritance**	Organelle inheritance	Defective ATP production affecting muscles.
Leber Hereditary Optic Neuropathy (LHON)	**Mitochondrial (Maternal) inheritance**	Organelle inheritance	Optic nerve degeneration → blindness.

High-Yield Exam Table (Disease → Inheritance)

Inheritance Pattern	Diseases Mentioned
X-linked (Atypical Repeat Expansion)	**Fragile X syndrome**
Mitochondrial (Maternal)	**Mitochondrial myopathy, Leber hereditary optic neuropathy**
Genomic Imprinting Disorders	**Prader–Willi syndrome, Angelman syndrome**
Locus Heterogeneity Example	**Congenital hearing loss (PAX3 AD, GJB2 AR)**
Allelic Heterogeneity Example	**Cystic fibrosis, Duchenne/Becker muscular dystrophy**
Pleiotropy Example	**Marfan syndrome, Cystic fibrosis**
Reduced Penetrance Example	**Retinoblastoma, Waardenburg syndrome**
Variable Expressivity Example	**Neurofibromatosis, Myotonic dystrophy**
Anticipation / Trinucleotide Repeat Diseases	**Fragile X, Huntington disease, Myotonic dystrophy, Spinocerebellar ataxia, Kennedy disease, Machado–Joseph disease, Friedreich ataxia**