Certainly! Below is a **concise, genetics-focused summary** of each disorder as per your professor's requirements, covering:
1. Genetic Defect | **2. Pathophysiology** | **3. Inheritance Pattern**

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1. Xeroderma Pigmentosum (XP)
- **Genetic Defect:**
  - Mutations in *XPA-XPG* genes (NER pathway).
  - **Types:** Missense, nonsense, or deletions disrupting DNA repair.
- **Pathophysiology:**
  - Defective NER → UV-induced pyrimidine dimers persist → DNA mutations → skin cancers.
- **Inheritance:** Autosomal recessive (25% recurrence risk if both parents are carriers).

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2. Osteogenesis Imperfecta (OI)
- **Genetic Defect:**
  - Mutations in *COL1A1*/*COL1A2* (glycine substitutions common).
  - **Types:** Null alleles (mild) vs. structural mutations (severe).
- **Pathophysiology:**
  - Abnormal collagen fibrils → brittle bones, fractures, blue sclerae.
- **Inheritance:**
  - Autosomal dominant (Types I–V) or recessive (Types VI–VIII).

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3. Cystic Fibrosis (CF)
- **Genetic Defect:**
  - **F508del** (3-bp deletion in *CFTR*) → misfolded protein (Class II).
  - Other classes: Nonsense (Class I), gating defects (Class III).
- **Pathophysiology:**
  - Loss of CFTR Cl^- channel → thick mucus → lung/pancreatic damage.
- **Inheritance:** Autosomal recessive (25% risk if both carriers).

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4. Retinoblastoma
- **Genetic Defect:**
  - Biallelic loss of *RB1* (tumor suppressor).
  - **Types:** Germline (40%) or somatic (60%) mutations.
- **Pathophysiology:**
  - Dysfunctional pRb → uncontrolled cell cycle → retinal tumors.
- **Inheritance:** Autosomal dominant (familial cases; 50% transmission).

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5. Huntington Disease (HD)
- **Genetic Defect:**
  - CAG repeat expansion (>36) in *HTT* (gain-of-function).
- **Pathophysiology:**
  - Toxic polyglutamine aggregates → neuronal death → chorea/dementia.
- **Inheritance:** Autosomal dominant (50% risk; anticipation if paternal transmission).

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6. Marfan Syndrome
- **Genetic Defect:**

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- **Genetic Defect:**
  - Mutations in *FBN1* (fibrillin-1) → dominant-negative effect.
- **Pathophysiology:**
  - Dysregulated TGF-β signaling → aortic aneurysms, lens dislocation.
- **Inheritance:** Autosomal dominant (50% risk).

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7. Hemophilia A
- **Genetic Defect:**
  - Mutations in *F8* (factor VIII): inversions (45%), deletions, nonsense.
- **Pathophysiology:**
  - Impaired clotting cascade → spontaneous bleeding (joints/muscles).
- **Inheritance:** X-linked recessive (50% risk to sons of carrier mothers).

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8. Duchenne Muscular Dystrophy (DMD)
- **Genetic Defect:**
  - Deletions (65%) or duplications in *DMD* → frameshift → no dystrophin.
- **Pathophysiology:**
  - Loss of dystrophin → muscle membrane instability → degeneration.
- **Inheritance:** X-linked recessive (50% risk to sons of carriers).

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9. Color Vision Deficiencies
- **Genetic Defect:**
  - Unequal crossover in *OPN1LW* (red) or *OPN1MW* (green) opsins.
- **Pathophysiology:**
  - Altered cone photopigments → red/green colorblindness.
- **Inheritance:** X-linked recessive (higher prevalence in males).

Key Patterns Summary Table
Disorder	**Gene**	**Genetic Defect**	**Inheritance**
XP	*XPA-XPG*	NER pathway mutations	Autosomal recessive
CF	*CFTR*	F508del (Class II)	Autosomal recessive
HD	*HTT*	CAG repeat expansion	Autosomal dominant
DMD	*DMD*	Deletions (frameshift)	X-linked recessive
Hemophilia A	*F8*	Inversions/nonsense	X-linked recessive

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10 Genetics-Focused MCQs
1. **XP results from mutations in:**
   a) Mismatch repair genes
   b) *XPA-XPG* (NER pathway)
   c) *BRCA1*
   Answer: b

2. **F508del in *CFTR* causes:**
   a) Hyperactive Cl^- channel
   b) Misfolded protein degradation
   c) Enhanced mRNA splicing
   Answer: b

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2. **F508del in *CFTR* causes:**
   a) Hyperactive Cl^- channel
   b) Misfolded protein degradation
   c) Enhanced mRNA splicing
   Answer: b

3. **HD's CAG repeats are in:**
   a) *HTT*
   b) *FMR1*
   c) *DMPK*
   Answer: a

4. **DMD patients lack:**
   a) Myosin
   b) Dystrophin
   c) Actin
   Answer: b

5. **Marfan Syndrome involves mutations in:**
   a) *COL1A1*
   b) *FBN1*
   c) *DMD*
   Answer: b

This summary **strictly adheres** to your professor's focus: **genetic defect → pathophysiology → inheritance**. Let me know if you'd like to adjust the depth for any disorder!