

Genetics Sickle Cell Disease and Hemophilia

From the Recordings to Notes

HCC Study Notes | Three Video Compilation

This guide condenses the three recordings into high-yield nursing notes. Focus first on inheritance patterns, hallmark findings, priority assessments, crisis management, medications, safety precautions, and patient teaching.

Rapid Review

Topic	Must Know
Genetics vs genomics	Genetics studies heredity and variation in inherited traits. Genomics examines the structure and function of the full genome and how DNA influences disease, prognosis, and treatment.
Sickle cell disease	Autosomal recessive hemoglobinopathy. Sickled red blood cells obstruct blood flow and break down early, causing ischemic pain, anemia, and organ complications.
Hemophilia	Usually X linked recessive. Hemophilia A is factor VIII deficiency; hemophilia B is factor IX deficiency. Prevent and rapidly control bleeding.
Top sickle cell priorities	Hydration, oxygenation when indicated, pain control, rest, infection evaluation and treatment, circulation checks, and early recognition of stroke or acute chest syndrome.
Top hemophilia priorities	Replace the missing factor, prevent injury, use bleeding precautions, apply RICE for bleeding, and monitor closely with transfusions.

Part One General Overview of Genetics

Why Genetics Matters in Nursing

- Genetic knowledge helps identify the hereditary components of disease.
- It can improve prediction of disease susceptibility, onset, progression, and response to treatment.
- Genetic disorders can affect anyone regardless of age, socioeconomic status, race, or ethnicity.

Genetics Compared With Genomics

Concept	Primary Focus	Clinical Relevance
Genetics	Mechanisms of heredity and variation in inherited traits or characteristics.	Helps explain how individual traits or disorders are passed through families.

Concept	Primary Focus	Clinical Relevance
Genomics	Structure, function, and evolution of all DNA and the mapping of genomes.	Connects the whole genome with disease development, prognosis, and treatment response.

Nursing Roles and Competencies

- Collect a complete family health history and use it to construct a pedigree.
- Incorporate genetic risk assessment into the plan of care.
- Give patients understandable genetic information, resources, and available services.
- Facilitate referrals to genetic counselors and other specialized services.
- Evaluate the impact and effectiveness of genetic and genomic technologies, interventions, and treatments.
- Participate in preconception counseling, prenatal screening, carrier testing, newborn screening, neonatal counseling, and palliative care for infants with life-threatening disorders.
- Provide specialized pregnancy care for patients with genetic conditions such as congenital heart disease, cystic fibrosis, and factor V Leiden.

Human Genome Project and Testing

- The Human Genome Project was an international effort to sequence and map all human genes, collectively called the human genome.
- The project was completed in April 2003 and contributed to advances in genetic testing.

Testing Area	Examples From the Recording
Genetic testing	Direct or molecular testing, linkage analysis, biochemical testing, and cytogenetic testing.
Prenatal testing	Maternal serum screening, fetal ultrasound, amniocentesis, and chorionic villus sampling.

Ethical legal and social considerations include protecting privacy and ensuring fair use and interpretation of genetic information.

Part Two Sickle Cell Disease

Core Concept and Pathophysiology

- Sickle cell disease is a group of inherited hemoglobinopathies in which normal adult hemoglobin is partly or completely replaced by abnormal hemoglobin S.
- Abnormal hemoglobin changes red blood cells into rigid sickle shapes. These cells do not flow freely and may obstruct small vessels.
- The two major problems are vaso-occlusion, which causes ischemia and infarction, and hemolysis, which causes chronic anemia.
- Clinical features may result from vaso-occlusion, vascular inflammation, increased red blood cell destruction, or a combination.

- Repeated sickling can make cells permanently sickled. Good hydration and oxygenation can reverse early sickling but cannot restore permanently damaged cells.
- Hemoglobin S red blood cells may survive only about 10 to 20 days, compared with about 120 days for normal red blood cells.

Inheritance and Epidemiology

- Inheritance is autosomal recessive: the child must inherit an affected gene from both parents to have sickle cell disease.
- When both parents carry the trait, each pregnancy has a 25 percent chance of an unaffected child, a 50 percent chance of a carrier with sickle cell trait, and a 25 percent chance of a child with sickle cell disease.
- Newborn screening is commonly used for early identification.
- The recording identifies sickle cell anemia as the most common form and notes higher prevalence among African American and Hispanic populations.
- It is a chronic illness. Children may be asymptomatic between crises and often participate in normal activities.
- Children younger than five are at greatest risk; overwhelming infection is a leading cause of death in this group.

Assessment and Diagnostic Focus

Assessment Area	What to Assess
History	Previous crises, usual triggers, effective home management, activity in the last 24 hours, fluid intake, temperature, exposure to illness, bleeding history, diet and iron intake, and frequent infections.
General and oxygenation	Fatigue, lack of energy, dyspnea, oxygen saturation, cyanosis, pallor, and whether the child is at baseline.
Pain and circulation	Pain location and pattern, especially joints and extremities; capillary refill; peripheral pulses; and extremity circulation.
Cardiac	S3 heart sound and murmurs.
Abdominal and genitourinary	Abdominal pain, hepatomegaly, splenomegaly, jaundice, hematuria, priapism, urine output, and proteinuria.
Neurologic	Behavior changes, loss of consciousness, seizure activity, vision changes, and early signs of stroke.
Diagnostics	CBC with differential, cultures when infection is suspected, chest x ray for respiratory concerns, kidney function tests, eye examinations, and transcranial Doppler screening for stroke risk.

Pain is patient-specific but often follows a similar pattern during each crisis. Do not assume every pain complaint is caused by sickle cell disease, and do not undertreat reported pain.

Major Sickle Cell Crises

Crisis	Cause	Expected Findings and Priorities
Vaso occlusive	Sickled cells clump in the microcirculation, producing ischemia and infarction.	Severe pain, swollen hands and feet, painful joints, abdominal pain, stroke symptoms, vision impairment, acute chest syndrome, jaundice, hematuria, or priapism. Restore circulation, hydrate, support oxygenation, and control pain.
Splenic sequestration	Blood pools in the spleen, causing rapid loss of circulating volume.	Profound anemia, hypovolemia, splenomegaly, hepatomegaly, circulatory collapse, and shock. Recurrent episodes may require splenectomy, which further increases infection risk.
Hyperhemolytic	Accelerated destruction of red blood cells.	Worsening anemia, jaundice, hematuria, reticulocytosis, and possible liver or kidney dysfunction. May require transfusion and organ support.
Aplastic	Bone marrow red blood cell production drops, often after viral infection or folic acid depletion.	Acute worsening of anemia with inadequate replacement of destroyed cells. Folic acid supplementation is commonly used for prevention and support.

Acute Chest Syndrome

- Symptoms may resemble pneumonia: chest pain, fever, shortness of breath, tachypnea, and cough.
- Treat respiratory symptoms as urgent because acute chest syndrome can become life-threatening.

Triggers to Prevent

- Fever or infection.
- Dehydration, especially during hot weather, illness, or decreased oral intake.
- Emotional or physical stress.
- Any condition that increases oxygen demand or interferes with oxygen transport.

Management During a Crisis

1. Assess airway, breathing, oxygenation, circulation, pain, neurologic status, and signs of infection or acute chest syndrome.
2. Promote hydration with oral fluids when tolerated and IV fluids when oral intake is inadequate. Replace electrolytes as ordered.
3. Administer oxygen when indicated and monitor pulse oximetry.
4. Provide prompt, adequate analgesia. The recording emphasizes that patients are often undermedicated.
5. Encourage bed rest to reduce energy and oxygen demand.

6. Obtain CBC with differential, blood and urine cultures, and chest x ray when fever or infection is suspected; administer antibiotics as ordered.
7. Administer blood products early when prescribed for severe anemia or to reduce ischemia.
8. Check extremity circulation at least hourly during an acute event, including capillary refill and peripheral pulses.
9. Keep extremities extended, avoid raising the knee section of the bed, and keep the head of bed at no more than 30 degrees as described in the recording to support circulation.
10. Avoid constrictive clothing. Keep the environment comfortably warm, not cold; the recording suggests approximately 72 degrees Fahrenheit.
11. Avoid unnecessary external compression. If a blood pressure cuff is required, use an unaffected extremity when possible.

Sickle Cell Medications and Therapies

Medication or Therapy	Purpose	Critical Nursing Teaching
Hydroxyurea	Increases fetal hemoglobin, decreases inflammation, reduces pain crises and acute chest syndrome, improves anemia, and may reduce transfusions and hospitalizations.	Take orally every day as prescribed. Monitor CBC closely because white blood cells or platelets may fall and anemia may rarely worsen. It is teratogenic; use reliable contraception during therapy and for the prescribed period after discontinuation.
Folic acid	Supports red blood cell production and helps prevent folate depletion associated with increased cell turnover.	Take consistently as prescribed.
Penicillin prophylaxis	Reduces serious bacterial infection risk, particularly in young children or those with functional asplenia.	Adhere to the regimen. Fever still requires prompt evaluation for sepsis and additional antibiotics may be ordered.
NSAIDs such as ibuprofen	May help manage early or mild pain when kidney function is adequate.	Hydrate and follow dosing instructions. Avoid or use cautiously when kidney problems are present.
Acetaminophen	Alternative pain medication, especially when kidney concerns make NSAIDs less desirable.	Use only within recommended total daily limits and consider other sources containing acetaminophen.
Opioids such as morphine or hydromorphone	Treat moderate to severe crisis pain; IV analgesia may be needed.	Monitor sedation, respirations, pain response, and safety. Meperidine or Demerol is avoided because its metabolite normeperidine can provoke seizures.
Blood transfusion	Treats severe anemia and may reduce ischemia when used early in selected complications.	Monitor according to transfusion policy and assess for reactions.

Long Term Complications

- Stroke, vision loss, acute chest syndrome, gallstones, splenomegaly or functional asplenia, hematuria and kidney damage, priapism, osteonecrosis of the shoulder or hip, osteomyelitis, chronic ulcers, abdominal pain, and hand foot syndrome.
- Repeated sickling may contribute to chronic anemia, impaired growth, delayed sexual maturation, and increased susceptibility to sepsis.

Patient and Family Teaching

- Recognize each child's usual early crisis signs and seek care promptly for uncontrolled pain, fever, breathing difficulty, neurologic changes, priapism, or reduced urine output.
- Prevent dehydration. Offer frequent water breaks during outdoor play and monitor for thirst, reduced urine output, or fewer wet diapers.
- Keep immunizations current, including pneumococcal, meningococcal, annual influenza, and recommended COVID vaccination.
- Use strong hand hygiene, reduce exposure to contagious illness, and avoid sharing personal items that spread infection.
- Use rest periods during heat or strenuous activity and avoid extreme temperatures.
- Keep regular follow-up for growth, weight, blood pressure, oxygen saturation, renal labs, eye examinations, and transcranial Doppler screening.
- Discuss inheritance and genetic counseling before future pregnancies.

Part Three Hemophilia

Core Concept and Types

- Hemophilia is a group of hereditary bleeding disorders caused by deficiency of specific clotting factors.
- Approximately 80 percent of cases discussed in the recording follow an X linked recessive inheritance pattern.

Disorder	Deficiency	Key Point
Hemophilia A	Factor VIII	Known as classic hemophilia.
Hemophilia B	Factor IX	Known as Christmas disease.
von Willebrand disease	von Willebrand factor with reduced factor VIII activity	May affect platelet adhesion as well as factor VIII stability.

Assessment and Laboratory Findings

- Assess for easy bruising and excessive or prolonged bleeding after minor cuts, bruises, abrasions, dental work, surgery, or trauma.
- Assess for bleeding into joints and muscles. Repeated hemarthrosis may cause chronic pain, loss of mobility, disability, and eventual joint replacement.
- Identify the specific deficient factor because treatment depends on the exact deficiency.

Test	Typical Hemophilia A or B Pattern
Activated partial thromboplastin time	Prolonged because the intrinsic clotting pathway is affected.
Prothrombin time	Usually normal.
Platelet count and platelet function	Usually normal in hemophilia A or B; abnormalities suggest another or additional disorder.
Bleeding time	Usually normal in hemophilia A or B; may be abnormal in von Willebrand disease.
Factor assay	Low factor VIII in hemophilia A or low factor IX in hemophilia B.
Fibrinogen	Assessed to help evaluate the broader coagulation picture.

Treatment and Medications

Treatment	Purpose	Nursing Considerations
Factor concentrate	Aggressively replaces the missing factor VIII or IX. Many patients learn home administration.	Give promptly for suspected bleeding according to the treatment plan. Confirm the correct factor and dose.
Desmopressin DDAVP	Can raise factor VIII activity about two to four times; used in selected mild factor VIII deficiency. May be given IV or intranasally.	Monitor response and fluid balance as ordered. It is not a replacement for factor IX in hemophilia B.
Aminocaproic acid	Antifibrinolytic that helps prevent breakdown of formed clots.	Administer as ordered and teach the patient its role in preserving the clot.
Packed red blood cells	May replace blood volume and red cells after significant blood loss.	Use an infusion pump and institution-specific blood tubing; monitor closely for transfusion reactions.
Platelets	Used only when specifically indicated by the clinical situation because hemophilia itself is a factor deficiency.	Verify the indication and follow transfusion monitoring standards.

Bleeding Prevention and Immediate Care

- Prevent injury by creating a safe home and activity environment and following individualized activity guidance.
- Use careful dental hygiene with a soft bristle toothbrush and gentle brushing to reduce gum trauma.
- Recognize and control bleeding quickly. For musculoskeletal bleeding, use RICE: rest, ice, compression, and elevation, while following the patient's factor replacement plan.
- Limit long-term joint damage through early treatment of joint bleeding and ongoing follow-up.

- Support the child and family with home-care education and genetic counseling when pregnancy is being considered.

Priority: prevention of bleeding comes first. After bleeding begins, replace the deficient factor promptly and protect the affected area.

Transfusion Safety and Apheresis

- Monitor for transfusion reactions using standard institutional guidelines.
- Use the appropriate infusion pump and specialized blood administration tubing rather than hanging blood to gravity.
- Use compatible fluids and follow facility policy before and during blood administration.
- Apheresis removes blood, separates its components, and reinfuses the remaining portion. It can be used to collect platelets from healthy donors.

High Yield Comparisons

Feature	Sickle Cell Disease	Hemophilia
Inheritance	Autosomal recessive.	Usually X linked recessive.
Primary problem	Abnormal hemoglobin causes sickling, vessel obstruction, hemolysis, and anemia.	Missing clotting factor causes impaired coagulation and prolonged bleeding.
Hallmark symptom	Pain from vaso-occlusion; chronic anemia is also common.	Prolonged bleeding and bleeding into joints or muscles.
Major emergency	Acute chest syndrome, stroke, sepsis, shock from sequestration, or severe anemia.	Uncontrolled internal or external bleeding, especially head injury or joint bleeding.
Core treatment	Hydration, oxygen when indicated, analgesia, infection management, hydroxyurea, and transfusion when needed.	Factor replacement, DDAVP for selected mild hemophilia A, antifibrinolytics, and injury prevention.
Key prevention	Avoid dehydration, infection, hypoxia, temperature extremes, and excess stress.	Avoid trauma and use bleeding and dental precautions.

Exam Focus Checklist

- Explain genetics versus genomics and identify core nursing competencies.
- Recognize the 25 percent affected, 50 percent carrier, and 25 percent unaffected pattern when both parents carry an autosomal recessive trait.
- Differentiate vaso occlusive, splenic sequestration, hyperhemolytic, and aplastic crises.
- Recognize acute chest syndrome and stroke as urgent sickle cell complications.
- Prioritize hydration, oxygenation when indicated, pain control, circulation assessment, and infection evaluation in a sickle cell crisis.
- Know hydroxyurea benefits, daily adherence, CBC monitoring, and teratogenic risk.
- Remember to avoid meperidine or Demerol in sickle cell pain because of seizure risk from normeperidine.

- Match hemophilia A with factor VIII and hemophilia B with factor IX.
- Recognize prolonged aPTT with usually normal PT and platelet count in hemophilia A or B.
- Prioritize factor replacement, injury prevention, bleeding precautions, and RICE for musculoskeletal bleeding.
- Know that DDAVP is used for selected mild factor VIII deficiency and aminocaproic acid prevents clot breakdown.

Memory Cues

- Sickle cell: hydrate, oxygenate when needed, medicate pain, and investigate fever.
- Hemophilia A equals eight; hemophilia B equals nine.
- Hemophilia affects the intrinsic pathway, so aPTT is prolonged.
- RICE protects a bleeding joint: rest, ice, compression, elevation.