

Fanconi Anaemia in Children: Bridging Genetic Complexity, Clinical Challenges, and Holistic Nursing Care

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ABSTRACT

Fanconi anaemia (FA) is a rare inherited multisystem disorder characterized by chromosomal instability, congenital abnormalities, progressive bone marrow failure, and increased susceptibility to malignancy. Common findings include short stature, abnormal skin pigmentation, upper-limb skeletal abnormalities, microcephaly, renal or genitourinary anomalies, and endocrine, ocular, auditory, or developmental problems. Bone-marrow failure often presents during childhood and may progress to pancytopenia. Diagnosis is supported by chromosome-breakage testing with diepoxy butane (DEB) or mitomycin C (MMC) and may be followed by molecular genetic testing. Management requires long-term haematology, genetics, and multidisciplinary surveillance. Hematopoietic stem cell transplantation (HSCT) is the only curative treatment for the hematologic manifestations, while other care is directed toward cytopenias, congenital anomalies, endocrine and nutritional needs, infection and bleeding prevention, and cancer surveillance. This journal-style review presents an illustrative paediatric case and emphasizes practical nursing assessment, family education, safety, and coordinated multidisciplinary care.

Keywords: Fanconi anaemia, pancytopenia, bone marrow failure, chromosomal fragility, paediatric nursing, HSCT, genetic counselling

INTRODUCTION:

Fanconi anaemia is a genetically heterogeneous disorder of DNA interstrand cross-link repair. Although the condition is named for its hematologic presentation, it affects multiple organ systems. Physical abnormalities occur in many affected individuals, but some children may have few or no obvious congenital features. Progressive cytopenias, especially thrombocytopenia or leukopenia, may be the first major clinical clue. Early recognition is important because affected children need specialized surveillance and treatment planning, and the diagnosis has implications for family testing and donor selection.^{1,5}

Case Scenario:

An 8-year-old girl is brought to a paediatric haematology clinic with increasing fatigue, easy bruising, recurrent minor infections, and reduced exercise tolerance for several months. Her parents report that she has always been shorter than peers. Examination

shows short stature, several café-au-lait-type pigmented macules, a small head circumference for age, and a hypoplastic thumb on one hand. There is no active bleeding. Her complete blood count shows anaemia, leukopenia/neutropenia, and thrombocytopenia. A bone-marrow examination shows marked hypocellularity. Because the combination of congenital findings and pancytopenia raises suspicion for an inherited bone-marrow-failure syndrome, chromosome-breakage testing with DEB/MMC is performed and shows increased chromosome breakage. Subsequent molecular testing identifies biallelic pathogenic variants in an FA-associated gene. The child and family are referred for multidisciplinary care and assessment for possible HSCT.

History And Initial Assessment:

- Presenting complaints: fatigue, easy bruising, recurrent infections, and reduced activity tolerance.

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- Growth/development: persistent short stature; school performance and developmental history should be assessed.
- Family history: ask about relatives with marrow failure, leukaemia, congenital anomalies, or unexplained early deaths.
- Physical assessment: skin pigmentation, limb/thumb abnormalities, head circumference, renal/genitourinary findings, hearing, vision, and endocrine features.
- Laboratory assessment: complete blood count with differential, reticulocyte count, marrow evaluation, and specialized chromosome-breakage/genetic testing as indicated.⁴

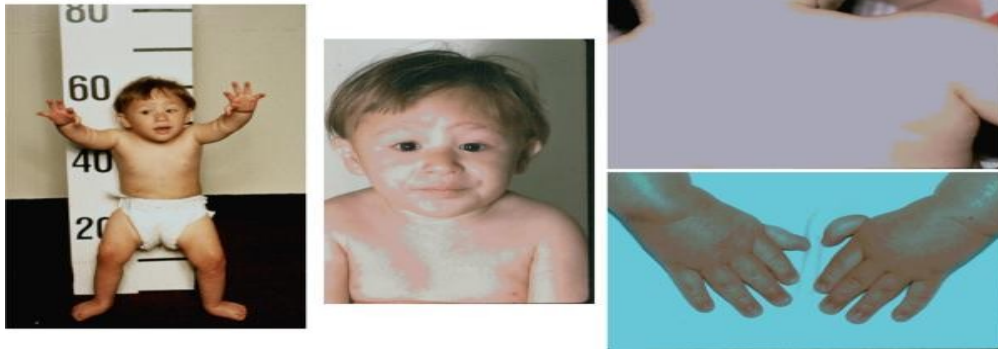


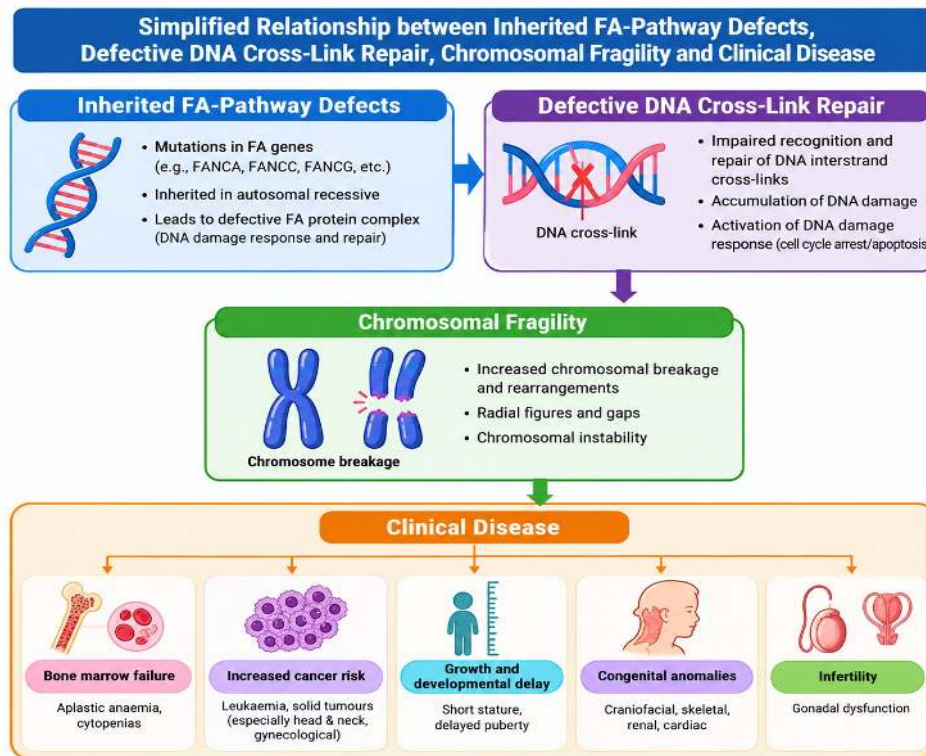
Figure 1: Overview of the History-Taking and Initial Assessment Process in Fanconi Anaemia, Including Patient and Family History, Clinical Evaluation, and Physical Examination.

Epidemiology:

Fanconi anaemia is rare. Orphanet reports an expected prevalence at birth of at least approximately 1 in 160,000 and classifies FA as a rare genetic multisystem disorder. The disorder is usually recognized in childhood, but age at diagnosis is variable because the physical phenotype can be subtle and some patients may initially present with bone-marrow failure rather than obvious congenital anomalies.¹³

Etiology And Genetics:

FA results from pathogenic variants affecting genes involved in the Fanconi DNA-repair pathway. Most forms follow autosomal-recessive inheritance; FANCB-related disease is X-linked, while RAD51-related FA can follow an autosomal-dominant pattern. Molecular testing is useful for confirming the genetic subtype, guiding family counselling, and facilitating testing of relatives and potential stem-cell donors.¹⁴



Flowchart: Simplified relationship between inherited FA-pathway defects, defective DNA cross-link repair, chromosomal fragility, and clinical disease.²

Clinical Manifestations:

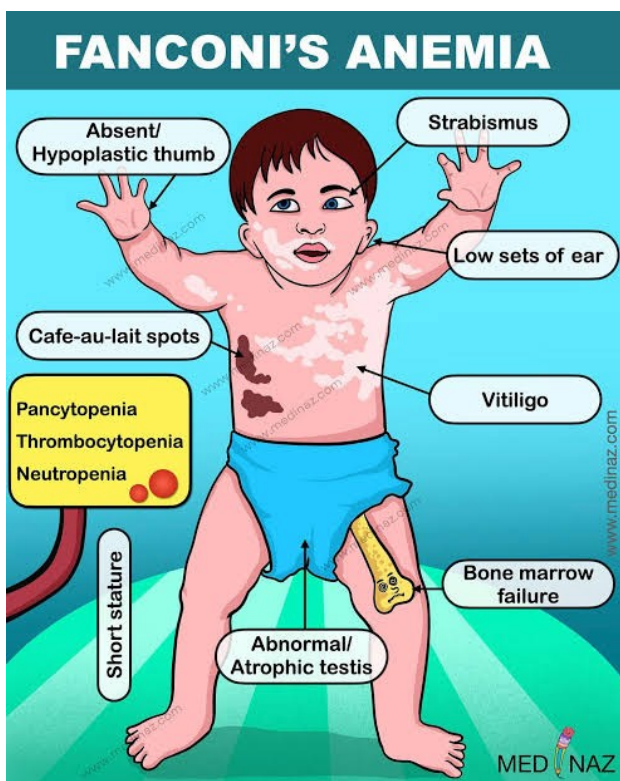
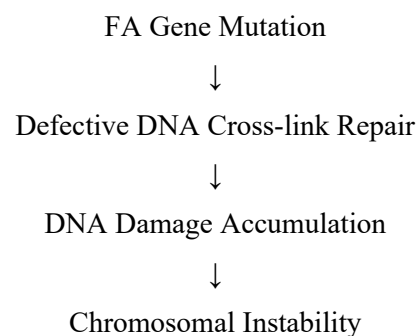


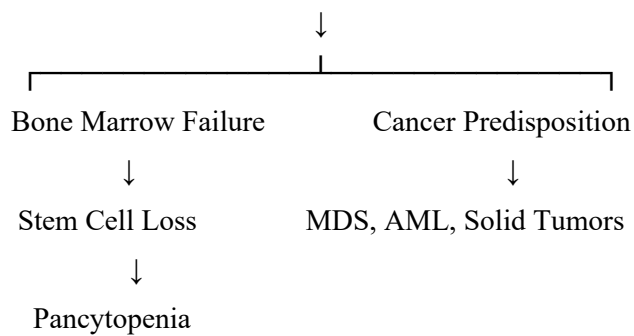
Figure 2: Clinical Manifestations of Fanconi Anemia, Including Congenital Abnormalities, Progressive Bone Marrow Failure, Hematologic Complications, and Increased Risk of Malignancies

Genetic Features:

- Autosomal-recessive FA: typically requires pathogenic variants in both copies of an FA-associated gene.
- X-linked FA: FANCB-associated disease follows an X-linked pattern.
- Autosomal-dominant FA: rare RAD51-related disease has been described.
- Genetic counselling is important for recurrence-risk discussion, family testing, and donor evaluation.
- A negative family history does not exclude FA because inheritance patterns vary and affected relatives may be undiagnosed.

Pathophysiology:





Common Findings

- **Hematologic:** progressive cytopenias, pancytopenia, aplastic marrow failure, MDS, and AML.
- **Skeletal:** hypoplastic or absent thumbs, radial abnormalities, scoliosis, and other limb differences.
- **Skin:** café-au-lait-type pigmentation or hypopigmentation.
- **Growth/endocrine:** short stature, thyroid abnormalities, glucose/insulin problems, delayed puberty, or gonadal dysfunction.
- **Renal/genitourinary:** structural urinary tract or genital anomalies.

Other: microcephaly, eye or hearing abnormalities, congenital heart defects, gastrointestinal anomalies, and developmental difficulties^{1,9}.

Diagnosis And Investigations:

Diagnosis begins with clinical suspicion and is confirmed with specialized laboratory testing. The current Gene Reviews chapter describes increased chromosome breakage and radial forms in lymphocytes after exposure to DEB and/or MMC as a key diagnostic finding. Molecular genetic testing can identify pathogenic variants and define the FA subtype.

1. Hematologic Assessment

- Complete blood count with differential and serial monitoring of blood-cell counts.
- Reticulocyte count and assessment for macrocytosis or elevated fetal haemoglobin when appropriate.
- Bone-marrow aspirate/biopsy for cellularity and morphology, with cytogenetic/FISH evaluation when indicated.

2. Chromosome-Breakage Testing

- DEB and/or MMC stress testing of lymphocytes is used to demonstrate characteristic chromosome fragility.

3. Molecular Genetic Testing

- Targeted FA gene testing, multigene panel, exome sequencing, or other molecular approaches may be selected according to the clinical situation.

4. Baseline Multisystem Evaluation

- Growth and endocrine assessment, including thyroid and metabolic evaluation.
- Renal/urinary tract imaging and assessment of congenital anomalies.
- Ophthalmologic and hearing evaluation.
- Cardiac evaluation when clinically indicated.
- Developmental, educational, nutritional, and psychosocial assessment.
- HLA typing of the child and relevant family members when HSCT is being considered⁷.

Differential Diagnosis:

The differential diagnosis includes other inherited bone-marrow-failure and chromosome-instability syndromes. Examples include dyskeratosis congenita, Diamond-Blackfan anemia, Shwachman-Diamond syndrome, Bloom syndrome, ataxia-telangiectasia, and Seckel syndrome. Molecular and chromosome-breakage testing helps distinguish FA from phenotypically overlapping conditions⁵.

Medical Management:

Management is individualized and should be coordinated through clinicians experienced in inherited bone-marrow-failure syndromes. Treatment addresses hematologic disease while monitoring non-hematologic manifestations throughout life.

Hematologic Care

- Supportive transfusion therapy may be required for clinically significant anaemia or thrombocytopenia, with careful attention to transfusion history and transplant planning.
- Androgens such as oxymetholone or other agents may improve blood counts in selected patients but require specialist monitoring for toxicity.

- G-CSF may improve neutrophil counts in selected patients under specialist supervision.
- HSCT is the only curative therapy for the hematologic manifestations of FA, although it does not eliminate the underlying non-hematologic and cancer risks¹¹.

Multisystem Care

- Endocrine, growth, renal, orthopaedic, hearing, ophthalmologic, dental/oral, nutritional, and developmental problems are managed according to individual findings.
- Cancer surveillance is an important component of long-term care because FA is associated with increased malignancy risk.
- Genetic counselling and psychosocial support should be integrated into the care plan.

Nursing Management:

1. Assessment of Fatigue and Anaemia

- Assess activity tolerance, pallor, dizziness, tachycardia, dyspnea, sleep pattern, and school participation.
- Monitor haemoglobin and other laboratory results as ordered.
- Cluster nursing activities and provide age-appropriate rest periods.
- Administer prescribed blood products or medications and monitor for adverse reactions.

Goal: Maintain adequate oxygenation, reduce fatigue, and support participation in age-appropriate activities.

2. Infection Prevention

- Monitor temperature and observe for subtle signs of infection, especially when neutropenia is present.
- Use meticulous hand hygiene and aseptic technique.
- Teach the family when to seek urgent medical attention for fever or infection symptoms.
- Follow the individualized immunization plan and specialist recommendations.

Goal: Prevent infection and identify infection early.

3. Bleeding Precautions

- Assess skin, gums, urine, stool, and other sites for bleeding or petechiae.
- Avoid unnecessary trauma and use gentle oral/skin care.
- Apply prolonged pressure after venipuncture when appropriate and follow platelet-related precautions.
- Educate caregivers about warning signs such as persistent nosebleeds, blood in urine/stool, or unusual bruising.

Goal: Minimize bleeding and prevent avoidable injury.

4. Nutrition and Growth

- Monitor weight, height, appetite, feeding difficulty, and growth trajectory.
- Provide small, frequent, nutrient-dense meals when appropriate and refer to a dietitian for individualized planning.
- Monitor for endocrine or gastrointestinal factors contributing to poor growth.

Goal: Support adequate nutrition and optimal growth.

5. Development, Schooling, and Psychosocial Support

- Assess developmental and educational needs and coordinate with school services.
- Encourage age-appropriate play and independence while balancing fatigue and medical restrictions.
- Provide emotional support and assess caregiver understanding, stress, and coping.

Goal: Promote developmental potential, participation, and family coping.

6. Preparation for HSCT When Indicated

- Explain procedures and expected phases using child-friendly language.
- Support HLA-related donor evaluation and required investigations.
- Monitor for infection, bleeding, nutritional problems, and treatment-related complications according to the transplant team's protocol.

- Provide family education about the need for long-term follow-up even after successful transplant.

Goal: Promote safe preparation, informed participation, and continuity of care.

7. Long-Term Surveillance and Health Education

- Reinforce scheduled haematology, genetics, endocrine, dental/oral, dermatology, hearing, vision, and other specialist visits.
- Teach sun protection and general skin/oral health measures as part of individualized cancer-prevention and counselling.
- Encourage families to keep a current medical summary and medication/transfusion history.

Goal: Improve early recognition of complications and adherence to lifelong surveillance.²

Multidisciplinary Care:

Children with Fanconi anaemia may require coordinated care from paediatric haematology, bone-marrow-transplant specialists, clinical genetics, endocrinology, nephrology/urology, orthopaedics, ophthalmology, audiology/ENT, dentistry/oral medicine, nutrition, developmental services, psychology/social work, and other specialists according to individual manifestations⁸.

Priority Nursing Problems

- Fatigue and activity intolerance related to anaemia.

- Risk for infection related to neutropenia or marrow failure.
- Risk for bleeding related to thrombocytopenia.
- Nutritional/growth problems when present.
- Knowledge deficit regarding disease, surveillance, medicines, and warning signs.
- Anxiety and altered family coping related to a chronic inherited disorder.

Family And Caregiver Education:

- Explain FA as an inherited disorder affecting DNA repair and blood-cell production using age-appropriate language.
- Teach the purpose of blood tests, marrow examinations, chromosome-breakage testing, and genetic testing.
- Teach caregivers to seek urgent medical advice for fever, unusual bleeding, severe weakness, breathing difficulty, or other symptoms identified by the treating team.
- Encourage adherence to scheduled surveillance because complications may occur even when the child feels well.
- Discuss genetic counselling, family testing, and donor evaluation when appropriate.
- Support school attendance and normal childhood activities as medically appropriate while respecting fatigue, infection, bleeding, and treatment-related precautions¹⁰.

Nursing Care Plan

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Evaluation
Fatigue, pallor, reduced activity, anaemia	Fatigue related to reduced oxygen-carrying capacity secondary to anaemia	Child will demonstrate improved activity tolerance and balanced rest/activity.	Assess fatigue; cluster care; Provide rest; Monitor CBC; Administer prescribed therapy/transfusion; Encourage gradual activity.	Fatigue reduced; child participates in age-appropriate activity.
Neutropenia/recurrent infections	Risk for infection related to neutropenia and impaired haematopoiesis	Child will remain free from preventable infection	Monitor temperature; Hand hygiene; Aseptic technique; Infection education; Follow individualized vaccination plan; Report fever promptly.	No avoidable infection; family demonstrates correct precautions.

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Evaluation
Thrombocytopenia, bruising/petechiae	Risk for bleeding related to thrombocytopenia	Child will remain free from significant bleeding or injury.	Inspect skin/mucosa; Reduce trauma; Gentle oral care; Monitor platelet count; Teach bleeding precautions; Follow transfusion orders.	No significant bleeding; caregivers verbalize precautions.
Short stature/poor growth or feeding concerns	Imbalanced nutrition: less than body requirements related to inadequate intake or disease-related needs, if present	Child will maintain adequate intake and follow an individualized growth plan	Monitor weight/height; Assess intake; small frequent meals; Dietitian referral; Manage contributing endocrine/GI problems.	Nutritional intake and growth trend improve or remain stable.
Child/family facing chronic inherited illness	Anxiety related to uncertainty, investigations, and long-term treatment	Child/family will verbalize understanding and demonstrate effective coping strategies	Use simple explanations; Allow questions; Provide psychological support involve family in care; connect with counselling/support resources.	Family demonstrates understanding and identifies coping resources.

Prognosis And Long-Term Follow-up:

The course of FA varies widely. Bone-marrow failure is progressive in many affected individuals, while some have later or milder hematologic manifestations. The risk of MDS/AML and solid tumors requires lifelong surveillance. HSCT can correct the hematologic manifestations of marrow failure but does not remove the underlying multisystem phenotype or all cancer risks. Long-term care therefore continues after transplantation¹².

CONCLUSION:

Fanconi anaemia is a rare inherited multisystem disorder in which defective DNA repair produces chromosomal instability, progressive bone-marrow failure, congenital abnormalities, and increased cancer susceptibility. Paediatric nurses have an important role in early recognition, ongoing assessment, infection and bleeding prevention, nutritional and developmental support, family education, and coordination of multidisciplinary follow-up. Accurate diagnosis also supports genetic counselling and safe planning for hematopoietic stem cell transplantation when indicated. Because FA remains a lifelong condition, nursing care should extend beyond the immediate hematologic problem to growth, endocrine, sensory, oral, psychosocial, and cancer-surveillance needs¹.

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