The Young Patient: Fanconi Anemia and Head and Neck Cancer

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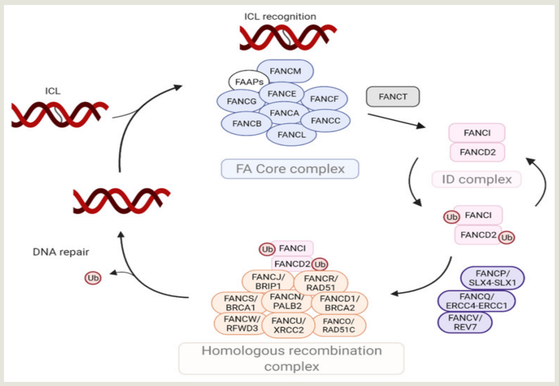
BACKGROUND

- Fanconi Anemia (FA) is a rare genetic disease that leads to defects in inter-strand DNA crosslink repair
- Leads to a 500-1000-fold increased risk of head and neck squamous cell carcinoma (HNSCC) compared to the general population
- Despite being younger at the onset of disease, FA patients tend to have worse survival rates - treatment options are limited
- FA patients lack the traditional risk factors associated with HNSCC
- Given rarity of FA, limited literature on FA-HNSCC exists
- This study aims to use a scoping review of the literature and a case presentation to describe the current understanding of FA and HNSCC, and update the literature that exists, which encompasses more remote cases

Pathophysiology of Fanconi Anemia



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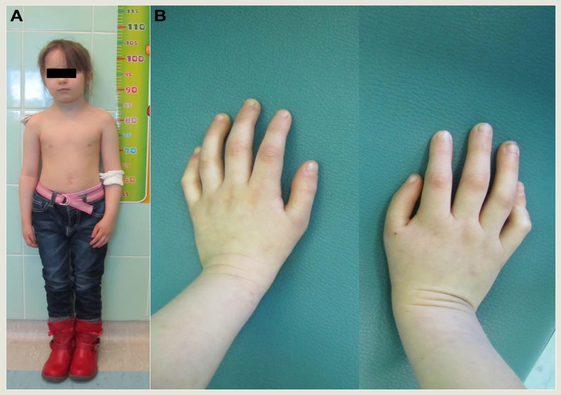
Classic Disease Characteristics



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STUDY DESIGN

- Scoping review using studies published between 2018-2022
- Inclusion criteria: case reports and case series of patients aged 12-80 with diagnoses of both FA and HNSCC
- Our case characteristics were combined with those from the literature
- Data extraction included: age of diagnosis of HNSCC, sex, TNM staging, presenting symptoms of HNSCC, history of stem cell transplantation therapy, primary cancer treatment, and whether HNSCC diagnosis preceded or followed FA diagnosis

FINDINGS

Characteristics	(N = 19)
Age (median)	32.5 (14 – 43)
Sex	
Male	11 (58%)
Female	8 (42%)
Smoking History	
Yes	1 (5%)
No	9 (47%)
Unknown	0 (47%)
Initial Presentation of Symptoms	
Tongue Lesion	9 (47%)
Other oral lesion	2 (11%)
Cervical nodule	1 (5%)
Dysphagia	2 (11%)
Foreign Body Sensation	1 (5%)
Odynophagia	2 (11%)
Hemoptysis	1 (5%)
Unknown	4 (21%)
T Stage	
T1	5 (26%)
T2	4 (21%)
T3	3 (16%)
T4	6 (32%)
Unknown	4 (21%)
History of Stem Cell Transplantation	
Yes	9 (47%)
No	7 (37%)
Unknown	3 (16%)
Treatment	
Primary Surgery Only	4 (21%)
Primary Surgery + adjuvant systemic therapy (XRT and/or chemo)	9 (47%)
Primary Systemic therapy (XRT and/or chemo)	6 (32%)
Chronicity	
FA diagnosis prior to HNSCC	15 (79%)
HNSCC prior to FA diagnosis	4 (21%)

- Our case: 28-year-old female with FA presenting with T4aN2bM0 oral cavity squamous cell carcinoma of the tongue
- Treated with primary surgical resection and adjuvant chemoradiation
- Treatment discontinued early due to chemotherapy related toxicity
- Nodal recurrence
- Patient died 2 months after treatment ended

CONCLUSIONS

Current studies show that patients with FA present with HNSCC at an earlier age than the general population. Due to the high incidence of oral cavity squamous cell carcinoma (OCSCC), **patients with FA should have regular cancer screening oral examinations. Diagnosis with HNSCC prior to 40 years of age or those with aggressive disease characteristics or extreme toxicities to treatment may warrant evaluation for FA.**

References

KU MEDICAL CENTER The University of Kansas