Abstract: PB3252

Title: EXPLORING THE LANDSCAPE OF MALIGNANCIES AMONG INDIVIDUALS WITH HEMOGLOBIN H DISEASE IN BRITISH COLUMBIA, CANADA

Abstract Type: Publication Only

Topic: Thalassemias

Background:

Alpha thalassemias are among the most prevalent blood disorders globally, with hemoglobin H (HbH) disease being the most common clinically significant form. Despite its prevalence, there is a paucity of data regarding long-term morbidities of HbH disease, including the incidence and pattern of both solid and hematologic malignancies. Only one other study from Italy has reported on these outcomes in HbH disease (Origa R et al. Cancer 2023).

Aims:

British Columbia, the western province of Canada, has a relatively large population with ethnic backgrounds from regions endemic for alpha thalassemia. This study aims to investigate the prevalence and pattern of malignancies in individuals with HbH disease in British Columbia.

Methods:

In British Columbia, all patients with clinically significant thalassemia, and the majority of patients with HbH disease, are referred to one of the two tertiary comprehensive hemoglobinopathy clinics at St. Paul Hospital or BC Children's Hospital. Patients with HbH disease were identified through The Inherited Coagulopathy and Hemoglobinopathy Information Portal (a data management tool for patients and clinicians in the BC Inherited Bleeding and Red Cell Disorders Services) or BC Children's Hospital hemoglobinopathy database. Patients' data were collected retrospectively using hospital based on provincial electronic medical record systems. We used The Surveillance, Epidemiology, and End Results (SEER) to classify cancer types. Data for the general population were obtained for comparison from the Canadian Cancer Society and Statistics Canada.

Results:

A total of 161 patients (90 females) with HbH disease were identified. The median age of the participants was 31 years (interquartile: 17-48; range: 1-94 years). Among the cohort, 144 patients had deletional HbH disease, 15 had non-deletional HbH disease, and 2 had homozygous alpha-gene mutations. Notably, six patients (3 females) were diagnosed with cancer. The key characteristics are summarized in Table 1. The ten-year incidence rate of cancer in individuals with HbH disease was 1.9%, in contrast to the 5.9% incidence rate reported for British Columbia with a median age of 41 years for the population. Of particular interest, one patient in the study developed hepatoblastoma at the age of 82 years. This patient, with no history of prior transfusions or hepatitis, had been on chelation for less than a year for a maximum liver iron concentration of 6.5 mg/gr dry weight. There was one cancer associated death during the study period. The predominance of females and the skewed age distribution towards younger ages are likely attributable to the identification of clinically asymptomatic patients during pregnancy and the recent establishment of provincial neonatal screening for hemoglobinopathy in British Columbia, respectively.

Case	Age at Dx	Sex	Genotype	Cancer Category	status	Regular Transfusion at Dx	Comments
1	29	F	Deletional	Breast	Recurrence at age 56. Remission	No	BRCA+
2	82	M	Deletional	Liver	Deceased	No	Hepatoblastoma
3	51	F	Deletional	Brain	Remission	No	Papillary tumor of the pineal region
4	68	M	Deletional	Prostate	Remission	No	Adenocarcinoma
5	32	M	Deletional	Testis	Remission	No	Testicular Seminoma
6	45	F	Deletional	Colon and Rectum	Remission	No	Adenocarcinoma

Summary/Conclusion:

In our cohort of patients, who were, on average, younger than the general population in British Columbia, the overall cancer incidence rate was not increased. However, one patient developed hepatoblastoma, a condition that is potentially associated with thalassemia-related complications, such as iron overload and transfusion-transmitted viral illnesses—an observation that was also reported in the Italian cohort. Regular surveillance screening for hepatoblastoma and active management of its risk factors are warranted in at-risk individuals with HbH disease, regardless of the genotype.

Keywords: Thalassemia, Malignancy