IHNS THE IRISH HEAD & NECK SOCIETY

THE JOURNAL OF THE IRISH HEAD AND NECK SOCIETY

Title: Insights into rare cancers: Characterizing the genomic landscape of anaplastic thyroid cancer

Body: Background: Anaplastic thyroid cancer is a rare and frequently lethal cancer, accounting for up to 2% of all thyroid cancer cases. Characterized by an aggressive phenotype and poor prognosis, systemic treatment options are limited. Recent advances in precision oncology have highlighted the importance of identifying actionable variants in rare cancers. Leveraging two independent datasets, we aimed to characterize the genomic landscape of this rare tumor type.
Methods: 229,453 samples from 196,244 patients available from AACR Project GENIE v.17.1 database were analyzed for the prevalence of mutations, fusions and copy number alterations in anaplastic thyroid cancer. Retrospective cases of anaplastic thyroid cancer were then assessed retrospectively in our institution from 2004-2022; clinicopathological information was collected and genomic analysis was performed to assess for most frequent actionable alterations.
Results: 324 samples were identified in 311 patients across 15 cancer centers. Anaplastic thyroid cancer occurred at a frequency of <1% of reported cancers (0.1%). 164 were identified as male (52%); 154 (49%) identified as female; 6 samples were unidentified. Median age was 67 (range 33-89). Most samples were taken from primary site 61% (n=200); versus metastatic site 31.5% (n= 99); and 7.7% (n=25) were unknown. Somatic mutations were noted in all 324 samples (100%). Most frequently altered genes included TP53 (58%), TERT (48.3%), and BRAF (40.1%), PIK3CA (15.7%)

Authors: Ruth Hutch, Stephen Finn, Michael Conroy, Cliona Grant, Niamh Coleman

Affiliations: St. James's Hospital,