

## English Abstract Session

2025年11月15日(土) 14:12 ~ 14:54 第10会場

**[E6] English Abstract Session 6 Miscellaneous**

Moderator:Kensuke Kumamoto(Department of Genome Medical Science and Medical Genetics, Faculty of Medicine, Kagawa University), Kamales Prasitvarakul(Hatyai Hospital)

### [E6-5] Clinicopathological and Genomic Characterization of Intestinal Adenosquamous Carcinoma

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**Background:** Adenosquamous carcinoma (ASC) is a rare malignancy with a poorly understood etiology. This study investigates primary intestinal ASC (IASC), with a specific focus on the ratio of these components and the underlying genetic alterations.

**Methods:** We retrospectively analyzed 14 patients diagnosed with IASC at Shanxi Cancer Hospital between 2012 to 2023 and the data on clinicopathological features were obtained. The proportions of adenocarcinoma and squamous cell carcinoma components were determined using immunohistochemistry (IHC). Whole-exome sequencing (WES) was performed on three primary duodenal ASC specimens and their corresponding normal tissues. This study was registered with the ChiCTR Registry (ChiCTR2400084845).

**Results:** The median patient age was 53 years, with a male predominance (64.3%). The primary tumor locations were duodenum (42.9%), jejunum (7.1%), right colon (21.5%), sigmoid colon (7.1%), and rectum (35.7%). All tumors contained both components exceeding 20% of the total volume, with 64.3% exhibiting a dominant squamous cell carcinoma component.

Human papillomavirus (HPV) infection and microsatellite instability (MSI) testing were negative for most patients. WES on three duodenal ASC cases revealed a predominance of missense mutations. Commonly mutated genes included SMYD4, NEB, SYNE1, and TP53BP1.

**Conclusion:** This study sheds light on the clinicopathological features of IASC and provides the first-ever whole-exome sequencing analysis of primary duodenal ASC. The identification of recurrently mutated genes in this aggressive malignancy presents promising targets for future therapeutic development. Further research is warranted to refine diagnostic strategies and explore targeted therapeutic approaches.