

## Introduction

- Malignant Melanoma of rectum is an extremely rare disease, aggressive in nature and often advanced at the time of diagnosis.
- It is mostly diagnosed in Caucasian women in fifth to sixth decade.
- We present a case of malignant melanoma of the rectum diagnosed on endoscopic polypectomy and staged with endoscopic ultrasound.

## Case Description

- A 66-year-old, asymptomatic, Caucasian female underwent surveillance colonoscopy.
- She was noted to have a 20 mm semi-pedunculated rectal polyp (fig. 1), which was piecemeal resected using a hot snare (fig. 2). Resection and retrieval were complete.
- Pathology showed malignant melanoma involving the anal and anorectal junction mucosa (fig. 3 and 4). Tumor approached the inked resection margin.
- Immunohistochemical stains were positive for Sox-10 (diffuse and strong) (fig. 5), Melan-A (focal), S-100 (focal), tyrosinase (focal). Stains were negative for pan keratin, CK7, CK20, CDX2, P40, CD56, Synaptophysin, CD45 and CEA.
- EUS performed 3 weeks later showed linear scar just above the dentate line with no residual polyp tissue. Rectal wall at the polypectomy site was thickened without any tumor infiltration and a normal appearing muscularis propria.
- Patient was noted to have two hypo echoic lymph nodes 11.4 mm X 8.5 mm and 6.4 mm X 6.0 mm, 8 cm from anal verge (fig. 6). FNA biopsy of the lymph nodes showed presence of malignant melanoma. Immunostaining was positive for MART-1 and SOX-10 but negative for HMB-45.
- PET/CT did not show any regional or distant metastatic disease.
- Tumor was staged as Stage III malignant melanoma and referred to tumor board.

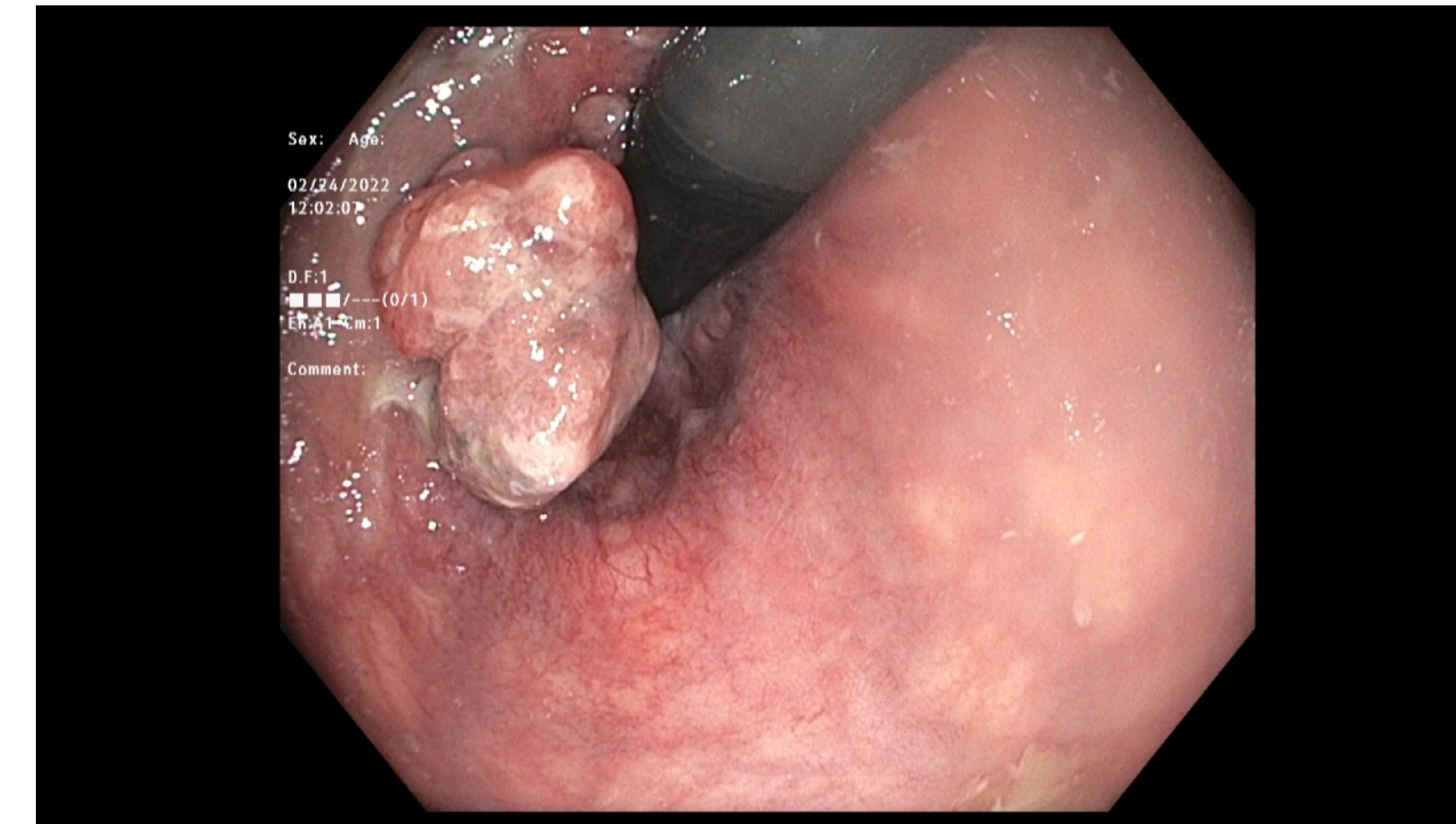


Figure 1

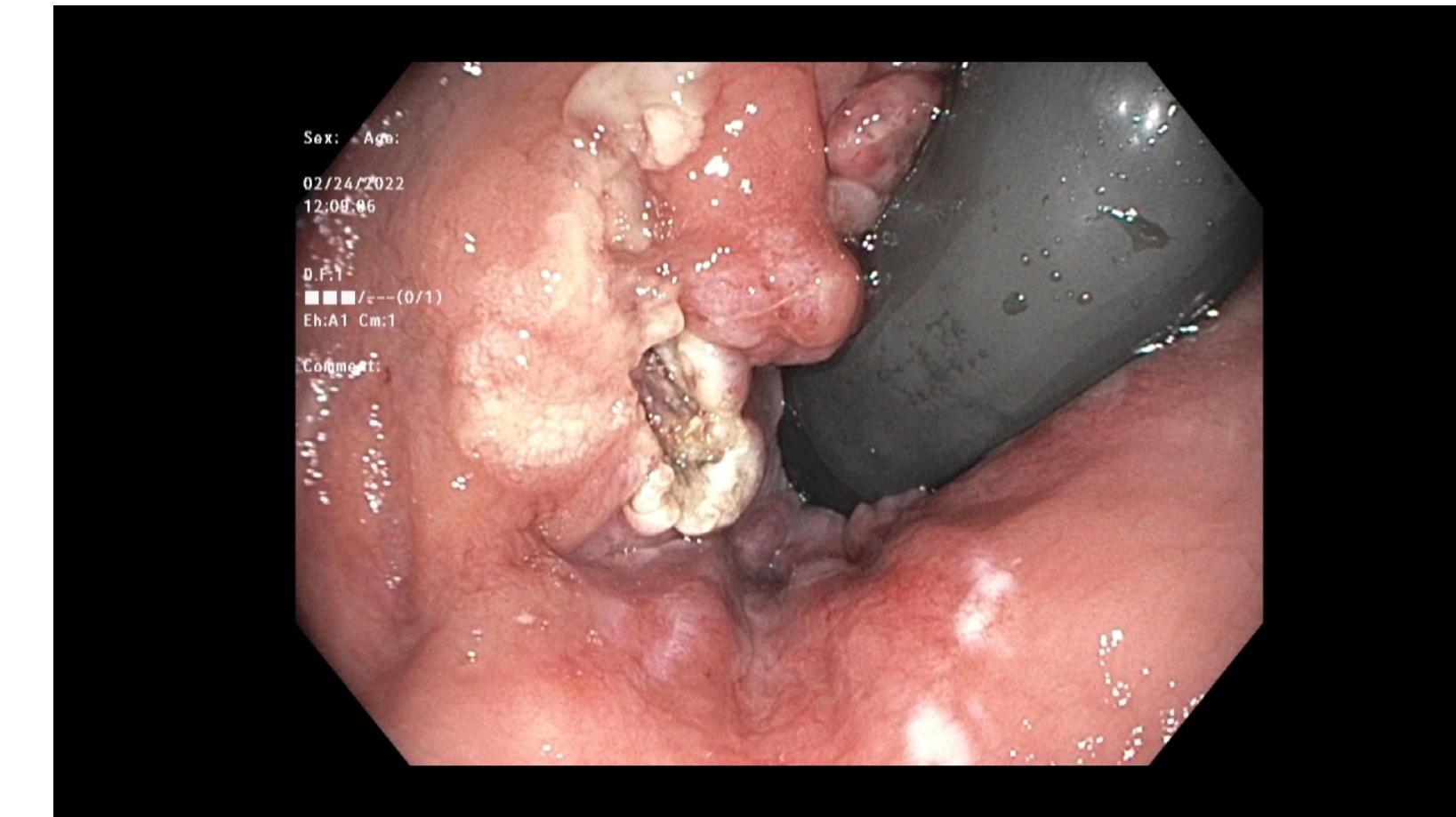


Figure 2

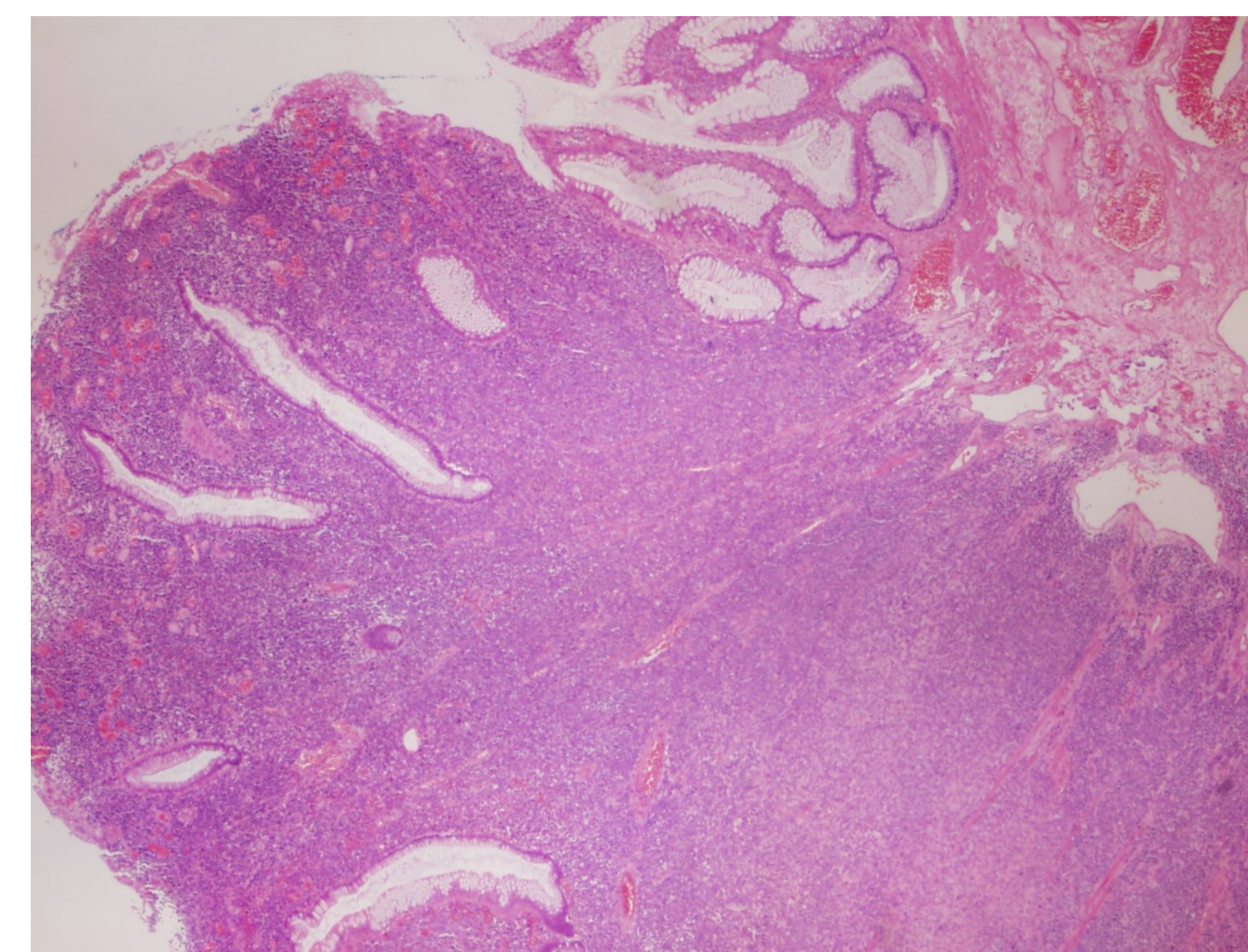


Figure 3

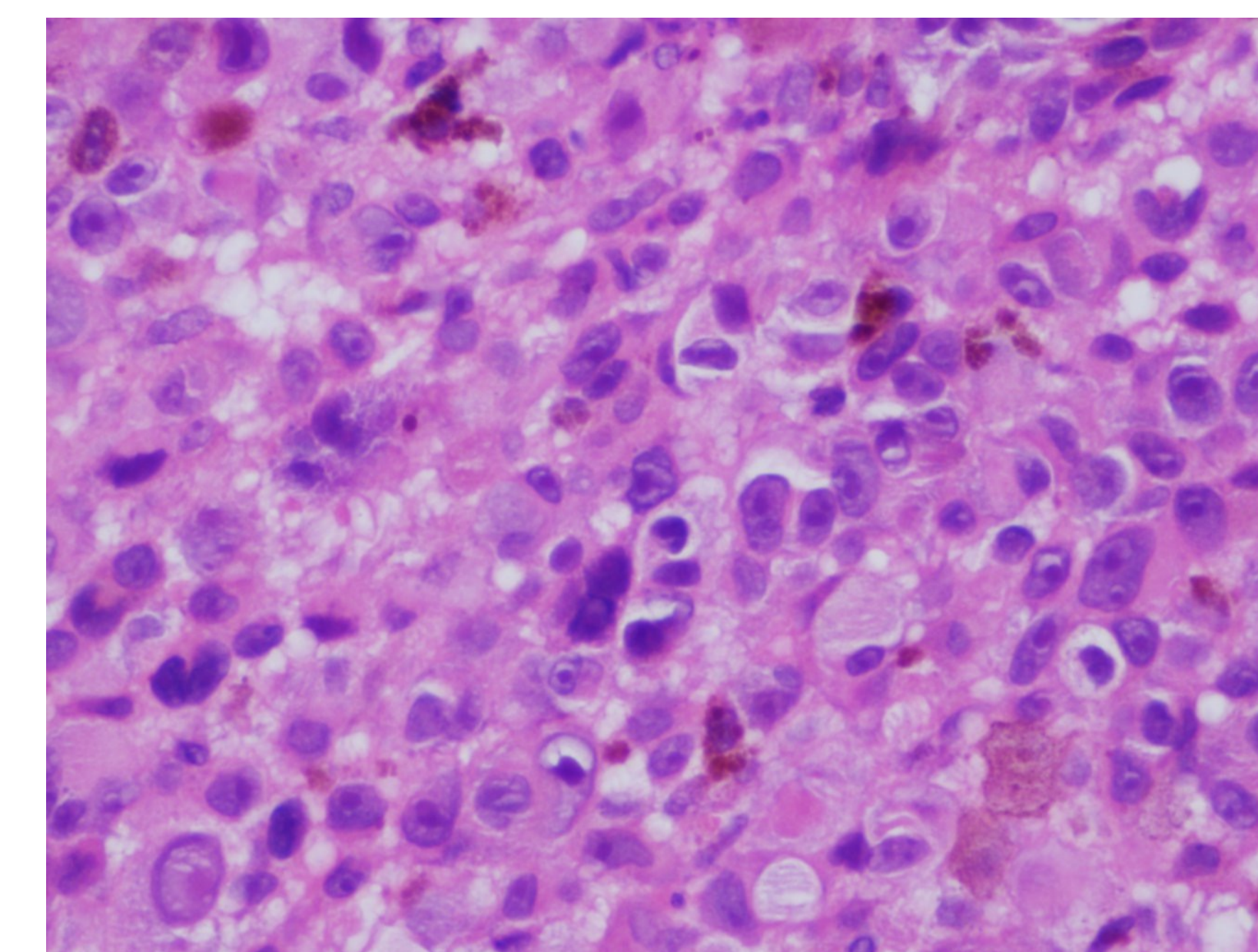


Figure 4

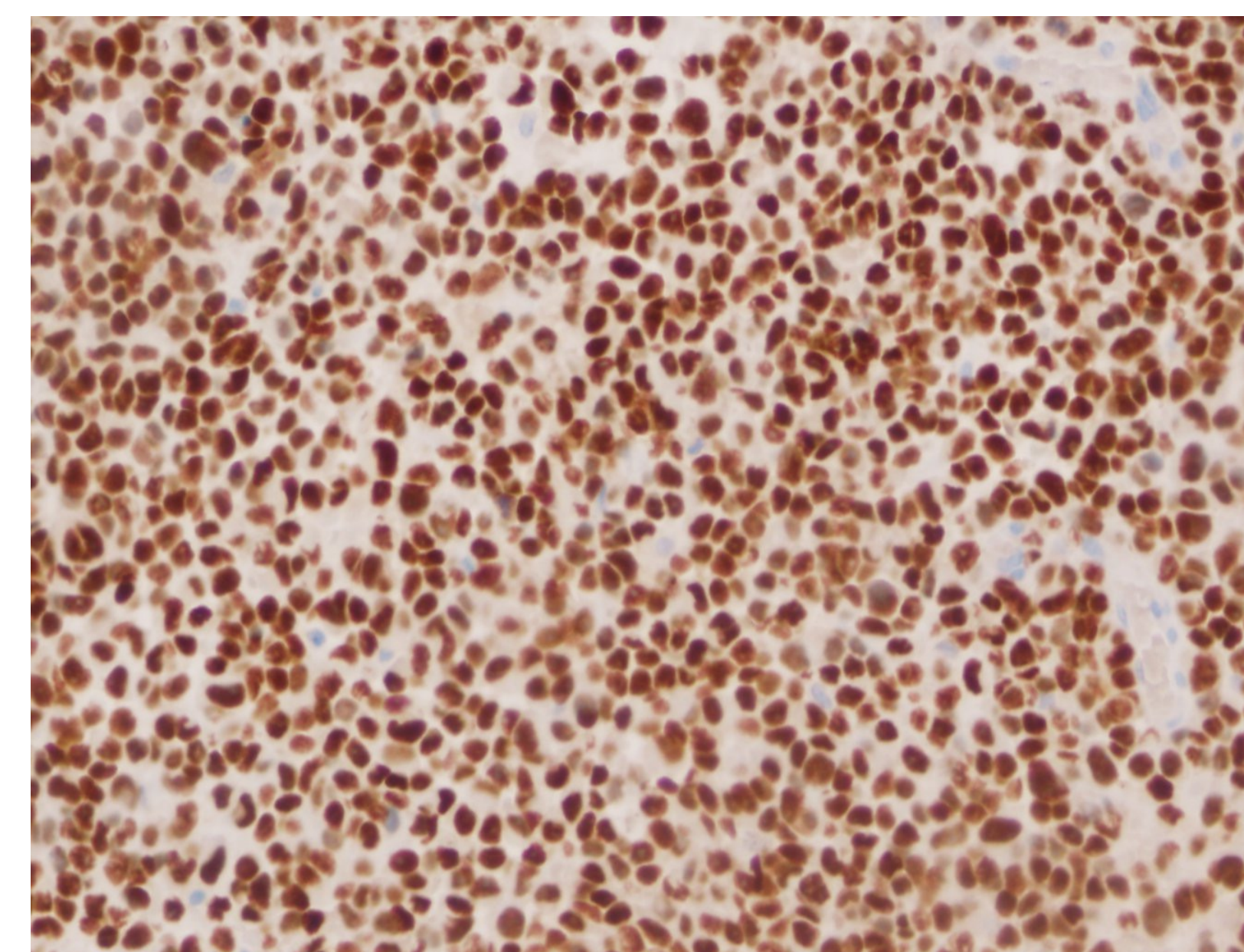


Figure 5



Figure 6

## Discussion

- Malignant melanoma of the distal rectum and anorectal junction is an aggressive and a rare presentation of this disease.
- Melanocytes are located at the anal transition zone and squamous zone with most anorectal melanomas arising from the dentate line and located at the anal verge or in the anal canal.
- Due to hidden location and late onset of symptoms many of these tumors are advanced by the time of diagnosis.
- As this case illustrates tumors are often 20 mm or bigger in size with nodal involvement.
- Hence, the five-year survival in patients with metastatic disease is only 16 percent.

## References

- Nederlandse Kankerregistratie (NKR) van IKNL Dataset Anorectaal Melanoom 2019
- van Schaik PM, Ernst MF, Meijer HA, Bosscha K: [Melanoma of the rectum: a rare entity](#). World J Gastroenterol. 2008, 14:1633-5.
- Reid A, Dettrick A, Oakenful C, Lambrianides A: [Primary rectal melanoma](#). J Surg Case Rep. 2011, 2011:2
- Huang, Wei-Feng, Xu Wang, and Wei Liu. "Primary Malignant Anorectal Melanoma." *Journal of Gastrointestinal Surgery* 26.1 (2022): 263-265.
- Garg, Vikas, et al. "Clinicopathological profile and outcomes of anorectal melanoma from a tertiary care center in India." *Future Science OA* 8.4 (2022): FSO786.