

Figure 1. HES coloration optical zoom x 50.

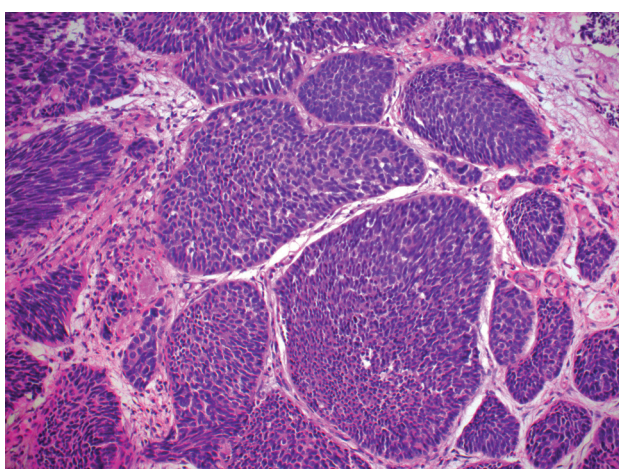


Figure 2. HES coloration optical zoom x 200.

treatment with an intravesical Bacillus Calmette-Guerin (BCG) therapy.

Due to the suspicion of the Gorlin Syndrome, the patient was also examined in the dermatologic department. The clinical examination showed a thoracic rash associated to angiomas and telangiectasias. Those lesions were also shown visible on the upper limb. The patient presented the characteristics of phototype I. He had no palmo-plantar pits, no signs of facial dysmorphism or skeletal abnormalities were noted.

Since the patient presented two major criteria of the Gorlin Syndrome, a genetic analysis of patched (PTCH) mutation had been realized but had showed no mutation of the gene. After a cystoscopic follow up of 5 years, the patient did not show any recurrence of the tumor in the bladder.

Discussion

Considering the assumption of the Gorlin Syndrome, we initially supposed this tumor could be an exceptional manifestation of the Gorlin Syndrome.

The Basal Cell Naevus Syndrome, also known as the “Gorlin Syndrome” is an inherited condition caused by a mutation in a tumor suppressor gene located on 9q22.3-31:

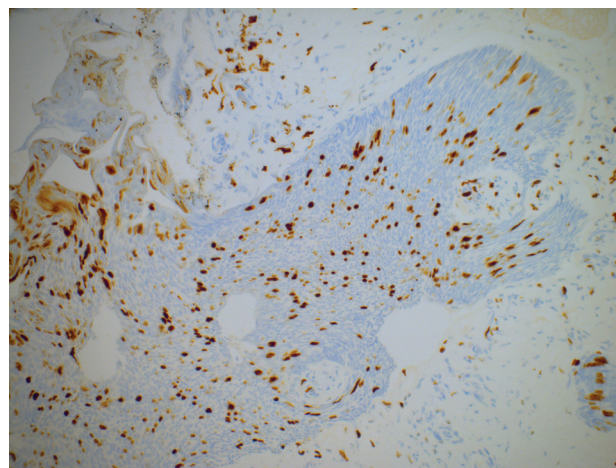


Figure 3. Ki67 labelling.

PTCH. The main clinical manifestations include multiple BCC's, odontogenic keratocytes, hyperkeratosis of palms and soles, skeletal abnormalities, ectopic intracranial calcifications, and facial dysmorphism.

The genetic justification for the suspicion of the bladder tumor as a manifestation of the Gorlin Syndrome was based on the fact that the chromosome 9 is the most rearranged chromosome in bladder cancer: from 50 up to 80% of bladder cancer cells show a loss of heterozygote of chromosome 9 [6]. Simoneau et al. [7] demonstrated that 9q22 region is a candidate for a tumor suppressor locus in bladder cancer. McGarvey et al. [8] detected two mutations in the PTCH gene among 54 invasive transitional cell carcinomas of bladder [8]. Moreover, Pignot et al. [9] observed under-expression of PTCH 1 gene in 31% of tumor samples of bladder cancer.

Even if our patient showed no mutation of PTCH gene, we could presume the possibility of a link between his skin tumors and his bladder tumor in reason of the similarity of their histology.

It would have probably been interesting to search in our patient for other mutations of genes involved in the pathogenesis of the tumor.

Conclusion

The basaloid carcinoma may be a rare variant of urinary bladder carcinoma, especially in patients with a medical history of recurrent Basal Cell Carcinoma of the skin.

What is new?

Few cases of basaloid squamous cell carcinoma have been described in bladder through some case reports. In this case, the patient presents a basaloid tumor, without any squamous differentiation. To our knowledge, it is the first case in the literature

List of Abbreviations

BCC	Basal cell carcinoma
PTCH	Patched
TUR	Transurethral resection

Funding

None.

Conflict of interests

The authors declare that there is no conflict of interests regarding the publication of this case report.

Consent for publication

Written informed consent was taken from the patient.

Ethical approval

Ethical approval is not required at our institution for publishing an anonymous case report.

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Summary of the case

1	Patient (gender, age)	Male, 61 years old
2	Final diagnosis	Basaloid carcinoma of bladder
3	Symptoms	Hematuria
4	Medications	BCG therapy
5	Clinical procedure	Transurethral resection of bladder tumor
6	Specialty	Urology, oncology