Poster 37: An exceptional case of an exceptional tumor: Porocarcinoma case report and recent updates.

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Background

Porocarcinoma (PC) is a rare adnexal tumor. The tumor arises from the acrosyringium of eccrine sweat glands. The risk of lymph node- and distant metastasis is high. Differential diagnosis with squamous cell carcinoma (SCC) is difficult, although NUT expression and YAP1 fusion products seem to be useful for diagnosis. Currently, wide local excision (WLE) is the main surgical treatment, although Mohs micrographic surgery is increasingly reported. To date there is no consensus regarding the role of sentinel lymph node biopsy (SNLB) and consequential lymph node dissection (LND). No guidelines exist for radiotherapy and systemic therapies.
We present a unique case of a pediatric porocarcinoma from our department. Furthermore, we report the findings of our narrative review we conducted on PC. A search string was used in PubMed and Embase, after which 21 articles were included in our review. We discuss epidemiology, clinical features, histopathological features, immunohistochemistry and fusion products, surgical management, radiotherapy, systemic treatments and outcomes.

Methods

For this narrative review, a search string was used in PubMed and Embase, from April first, 2012, until February twenty-sixth of 2023. The manual screening process of the results is represented in figure 1.

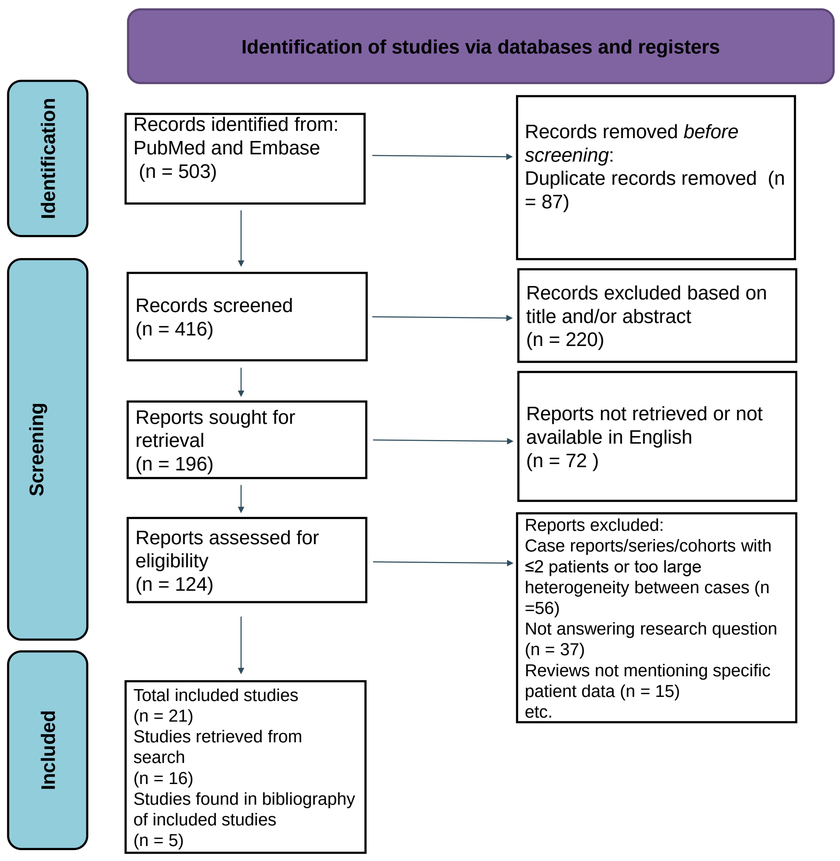

Figure 1. Visual representation of the search process conducted

Case presentation

History:
14-year old male
Pedunculated bleeding nodule on the scalp
Present since 3-4 months

Differential diagnosis:
Dermal nevus
Spitz nevus
Pyogenic granuloma

Management:
Shave biopsy

Histology:
Nuclear atypia
Mitotic rate +++
Duct formation

Immunohistochemistry:
NUT-positive
YAP1-NUTM1 fusion

Diagnosis:
Eccrine porocarcinoma

Staging:
PET-CT: negative
Brain MRI: negative

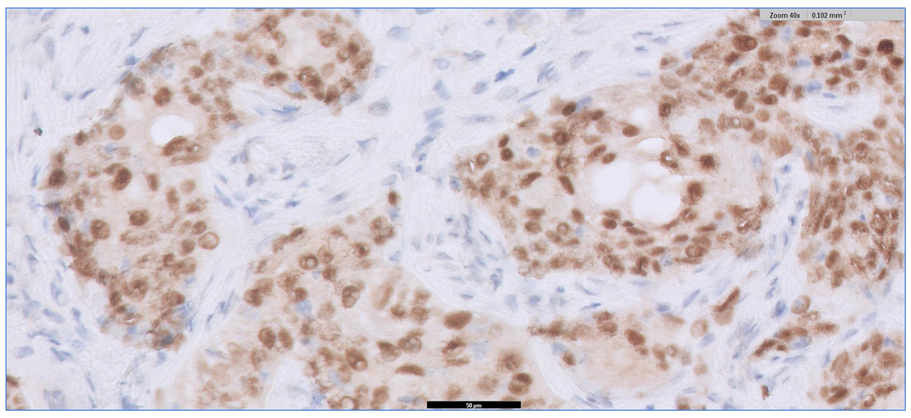

Figure 2: NUT-staining surrounding ductal structures in our patient

Results

- Our literature search yielded 503 results. After duplicate analysis and article selection (figure 1) we included 21 studies.
- Porocarcinoma is an **extremely rare adnexal tumor** mainly found in the 6th to 8th decades. Only few case reports on pediatric PC exist.
- **Main clinical presentation** is an erythematous nodule most frequently found on the head- and neck region, followed by the lower limbs.
- A large meta-analysis reported 453 PC and report a **metastasis rate of 19.8% and 14.5% for lymph node- and distant metastasis**, respectively. Most frequent areas for distant metastasis were lung (12.8%), liver (9%), brain (9%) and skin (5.8%)
- **Diagnosis can be challenging**, since PC can mimic SCC clinically and histologically. Mean delay to diagnosis is >5 years.
- NUT expression is specific for poroid neoplasms but not very sensitive for PC. **YAP1 fusion products, either YAP1-NUTM1 or YAP1-MAML2, are present in around 70% of PC** and are also specific for poroid neoplasms. In our case the presence of NUT expression and YAP1 fusion products was assessed since histology was suggestive for PC. The diagnosis of PC could be confirmed by demonstrating NUT positivity (figure 2) in combination with a YAP1-NUTM1 (exon 3) fusion product. It is hypothesized these fusion products could also play a pathogenic role.
- The main surgical treatment is WLE with margins of 1-3 cm**. Mohs micrographic surgery seems promising** in reducing risk of local recurrence, regional recurrence and distant metastasis but further studies are needed. In our patient malignancy was not suspected based on clinical presentation, so initial shave biopsy was performed. After negative PET-CT and MRI of the brain, WLE with a margin of 1 cm was planned. Unfortunately, no residual scar could be found, hence WLE was not possible. The patient was planned for careful monthly clinical follow-up and MRI every 6 months for assessing local invasion.
- **The role of SNLB and LND in PC is unclear**. There are no prospective comparative studies for treatment of nodal disease.
- No clear indications exist for the use of radiotherapy. **Main reasons for the use of radiotherapy** are tumor characteristics, margin status and tumor localization.
- Reports of chemotherapy are very heterogenic. **Most frequent chemotherapeutics used are carboplatin and cisplatin**. Treatment with **pembrolizumab** is reported in 3 cases and seems promising.
- One large cohort reported excellent **5-year disease specific survival rates** in localized disease (97,4% if tumor <2cm) but substantially worse survival rates in case of nodal involvement or deep dermal invasion (66%).

Conclusion

We conclude that PC is a very rare cutaneous tumor mainly occurring in the elderly in the head- and neck regions and lower limbs. Diagnostic delay remains an important problem. NUT expression and YAP1 fusion products may facilitate the diagnosis since they are specific for poroid neoplasms, but combination with histology is necessary. The risk for both nodal- and distant metastasis is high. Mohs micrographic surgery seems promising as surgical treatment. Further studies are needed regarding benefits and indications of SNLB, LND, radiotherapy and systemic therapies. We suggest that all cases of PC are referred to tertiary care hospitals so prospective studies can be conducted. For staging we recommend PET-CT and MRI/CT of the brain. Surgical treatment, sentinel lymph node biopsy and adjuvant therapies should be discussed multidisciplinary.

References

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