Merkel Cell Carcinoma; A case of a rare disease

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Introduction:
Merkel cell carcinoma (MCC), also known as neuroendocrine carcinoma, is a rare disease with around 1,500 cases per year in the United States. Because MCC is often fatal, diagnosis and immediate treatment are necessary. MCC usually presents in sun-exposed areas, with a small, painless, red-blue colored papule which grows rapidly over weeks and months, with the ability to metastasize. Risk factors include fair skin, sun exposure, age over 65, female gender, and chronic immunosuppression. MCC arises from highly atypical granules, that are noted to have similar structure and histological findings to those with neuronal and hormonal function. Although the exact cause of the carcinoma is unclear, recent studies have found a possible link between a polyomavirus found in MCC tissue as a possible cause of the disease. Treatment is determined by the progression of disease at time of diagnosis. Complete surgical excision of the lesion, followed by sentinel lymph node biopsy is the initial treatment. The need for radiation therapy or chemotherapy, are determined on a case by case basis.

Case Study:
My patient was a 90 year old Hispanic female who presented to clinic with a 3 month history of a tender, growing lesion to her left upper extremity. The patient had a past medical history which included diabetes and hypertension, both controlled by medications and followed by her PMD. She denied any fever, weight loss, lachrymography or personal/family history of skin cancers. Physical exam showed a red-violaceous “juicy” appearing round nodular plaque to the posterior aspect of the patient’s left upper arm. No other lesions were noted throughout the rest of the physical exam. There was no lymphadenopathy noted. A shave biopsy of the lesion was done, which showed histology consistent with a Merkel cell carcinoma. Pt was referred to an oncologist, and ultimately lost to follow up.

Discussion:
Epidemiology:
Most commonly presents in the elderly population, with an increased female to male ratio. Other risk factors include UV-exposure and fair skin.

Pathogenesis:
MCC is recognized as being a “primary neuroendocrine carcinoma of the skin.” Due to the similarities noted in this tumor with normal Merkel cell findings (as well as other neuroendocrine cells). The Merkel cell, is found in the basal layer of the epidermis and is recognized as a receptor touch cell. Recent studies have shown an association between Merkel cell carcinoma and a Merkel cell polyomavirus (MCV). 80% of patients noted to have MCC; are found to carry the virus, and evidence shows that the MCV plays a causative role in most MCC. The remainder of cases of MCC without the presence of the MCV, have a different etiology.

Clinical Manifestations:
Although most often found on the head and neck, MCC can also be found in the extremities and buttocks. It most commonly presents as a pink-red to violaceous, firm, dome shaped nodule with a rapid growth cycle. There is a high risk of recurrence after excision of the lesion. The size of the tumor is a determining factor in patient prognosis; lesions >2 cm have a 51% 5-year survival rate, whereas those lesions <2 cm have a 5-year survival rate of 66%. Lymph node involvement and metastasis further lower the patients’ overall 5-year survival rate.

Diagnosis:
Initial diagnosis is done by biopsy of the skin lesion in question. Once the lesion is biopsied, it is looked at under a microscope for distinguishing features specific to a Merkel cell carcinoma. Biopsy of a portion, or the entire lesion, is done. In order to determine if there is any spread of cancer cells, a PET or CT scan of the body is done in patients with lesions >2 cm, and with symptoms suggestive of lymph node involvement. Sentinel node biopsy may be done to determine lymph node involvement, and progression of spread.

Pathology:
The MCC tumor presents as a poorly defined mass, noted in the dermis. The mass often infiltrates into the subcutaneous fat, fascia and muscle. The growth pattern is composed of monomorphic small blue cells with small amounts of cytoplasm. Nuclear molding, apoptosis, and mitosis are often seen in these cells. Staining for specific neuroendocrine markers, as well as immunohistochemical findings further aid in the diagnosis of MCC. CK20 (a low molecular weight cytokeratin) often stains positive in a perinuclear globule pattern, and markers such as synaptophysin, chromogranin, and neuron-specific enolase, stain positive as well. PCR can be employed to determine the presence of Merkel cell polyomavirus, however false-positive PCR are commonly seen. The presence of p63 expression further shows an increase to the aggressive nature of the tumor.

Treatment:
Initial treatment in MCC is surgical excision. Depending on the tumor stage, radiation therapy and chemotherapy may be indicated. The presence of palpable lymph nodes on physical exam, indicate the need for biopsy of the node. Whereas, non-palpable lymph nodes on exam indicate the needs for wide-local excision with sentinel lymph node biopsy, and further treatment dependant on sentinel node biopsy results.

Conclusion:
Although a rare disease, the immediate identification of a Merkel Cell Carcinoma is of extreme importance in providing treatment and improving patient prognosis. While the exact cause is unknown, further investigation into a viral etiology is of interest in finding new treatment modalities, and provides new insight into a possible viral-oncologic relationship.

Images:

Pathology:

- Small blue cells with minimal amounts of cytoplasm
- Tightly packed nuclei in sheets
- Apoptosis, mitosis also seen
- Usually localized to dermis
- Immunohistochemical stains positive for CK20, CD56, chromogranin, synaptophysin, neurofilament

References:

