Rare type of masquerade uveitis. Case report.

Background: Uvea is an extremely rare localization of rhabdomyosarcoma (RMS). There are only three cases reported in the literature.

It accounts about 5% of pediatric cancer and can occur in any site, including the ocular region (20% of all RMS, mostly orbital). RMS arises from skeletal or striated muscle but there are no such muscles present in the uvea. Its pathogenesis is unclear.

Localization:

Head and neck 45% (30% arise in the orbit), trunk 40% and extremities 15%.

Pathology:

1. Pleomorphic (adults)
2. Embryonal (infants, head and neck, the majority of orbital RMS)
3. Alveolar (Children, most malignant, head and neck, less common in orbital region)
4. Bortyoid (very rare)

Can be primary, secondary and metastatic – scalp, face, buccal mucosa, oropharyngs, laryngs.

Case report.

5 yo Norwegian boy with a granulomatous iritis and secondary glaucoma in his left eye from June 2013.

Clinical findings did not confirm any infections, autoimmune pathology or tumors such as lymphoma, leukemia or retinoblastoma.

Radiological findings:

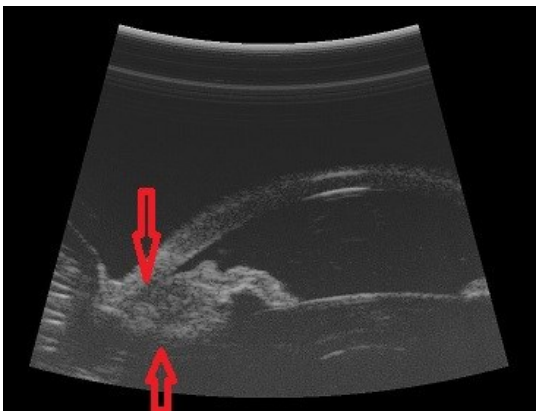
No tumors or foreign bodies identified by ultrasound and MRI of cerebral and orbital region.

Clinical challenges:

Because of high ACE, he was referred to the pediatric department where Blau syndrome was excluded.

His iridocyclitis and glaucoma did not respond to any treatment. Trabeculectomy and subsequent Ahmed Valve were performed without effect.

When the precipitate pattern grew confluent, a new ultrasound was performed with a suspected slightly thickening of the ciliary body and iris around the Ahmed Valve with a white mass in the angle of the anterior chamber seen by gonioscopy. His eye (Visual acuity 0,5 Log MAR) was enucleated due to a strong suspicion of malignancy**.**



The tumor of the cilary body had a size of 2,5mm x 3mm. It infiltrated an area around Ahmed valve which lead to an occluded angle from 12:30 to 2 o'clock.

Histology:

Rhabdomyosarcoma of ciliary body of alveolar (most malignant) type.

Management.

The patient received radiotherapy and chemotherapy. No local or distant metastasis were occurred.

Discussion:

RMS of the ocular region is an uncommon localization of this tumor. This malignant neoplasm develops from striated muscle in various stages of embryogenesis.

Conclusion:

Our workup took 9 months. What could we have done different to recognize a tumor with such a difficult localization in an earlier stage to be able to start a lifesaving treatment?

We should never trust ourselves to a single anterior chamber tap without detected malignant cells in the aqueous. In cases of uveitic non-responders to any therapy it should be repeated with a consideration of malignancy.

Gonioscopy repeated with a 2 month interval could help to get a better control over the iris back surface and relations in the angle. It could be difficult to perform in children. We have not used it enough in this case – only initially and in the final stage when the patient was under general anesthesia.

The resolution and frequency of the ultrasound plays an important role in the diagnosis of such a situated tumor where other means of examinations are difficult.

Lymphoma, leukemia, retinoblastoma and metastatic tumors are not the ONLY tumors of masquerade uveitis. Medulloepithelioma, juvenile xanthogranuloma and melanoma are also rare malignant disorders manifesting as uveitic glaucoma in the pediatric group. We probably do not find RMS associated uveitis within any of the manuals.

References:

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