

# VIRCHOWS ARCHIV

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Abstracts

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50 years (range, 18–80 years), with no gender predilection. The differential diagnosis of conjunctival myxoma includes amelanotic melanoma, fibrous histiocytoma, lymphangioma, amelanotic nevus, lymphoma, myxoid neurofibroma, spindle cell lymphoma or rhabdomyosarcoma. Conjunctival myxoma can be misdiagnosed as a conjunctival cyst. Conjunctival myxomas can occur in association with the Carney Complex, which is an autosomal dominant syndrome associated with benign tumours, spotty mucocutaneous pigmentation, and endocrine overactivity.

#### PS-11-011

##### **Orbital cystic schwannoma arising from optic nerve**

Z. Bayramoglu\*, E. Ayik, C. I. Bassorgun, G. O. Elpek

\*Akdeniz University, School of Medicine, Pathology, Antalya, Turkey

**Objective:** Most of the primary tumours of optic nerve are meningiomas and gliomas. Schwannomas arising from optic nerve are extremely rare lesions and only a few case has been reported until today. These lesions are slow-growing, well-circumscribed, solid/cystic masses. Here we reported a rare lesion schwannoma located in optic nerve with a cystic morphology.

**Method:** case report

**Results:** A 57-year-old female patient presented with a left ocular pain and progressive left proptosis. The patient was otherwise healthy. Orbital magnetic resonance imaging revealed an intraconal heterogeneous mass of 25 mm × 18 mm, radiographically consistent with a cavernous hemangioma. Surgical resection was performed and according to histopathological and immunohistochemical examination the final diagnosis was determined as "Orbital Schwannoma with Cystic Morphology".

**Conclusion:** Peripheral nerve tumours comprises 2 % of all orbital tumours and schwannomas are the most common types. Schwannomas are benign tumours arising from Schwann cells and most commonly involve trigeminal nerve root in intracranial location. Even if it is rare, schwannomas arising from optic nerve can be seen and cystic morphology of these tumours should not mislead the diagnosis.

#### PS-11-012

##### **Keratoplasty in patients with Acanthamoeba keratitis: A study of three cases**

L. Alfaro\*, C. Peris-Martinez, M. Roca-Estelles

\*Valencia, Spain

**Objective:** Corneal infections by amoebas present difficulties for clinical diagnosis and may be confused with other keratitis of the herpetic, fungal or bacterial type. We reviewed the clinical features and evolution in three patients with amoebic keratitis who required corneal transplantation after failure of medical treatment.

**Method:** The studied samples were three specimens of penetrating keratoplasty. Patients were two women aged 22 and 28 years and a 40-year-old male. One patient underwent a small corneal biopsy 1 week before transplantation, which was negative for amoebae. Samples were processed in a conventional manner and in addition to H/E sections Trichrome, PAS and Gram techniques were performed.

**Results:** The three patients revealed corneal cysts and trophozoites of Acanthamoeba. The epithelium was detached in all three although only two presented significant ulceration. One of the patients who had developed a crystalline keratopathy had superficial stromal band deposits interpreted by accumulation of the drugs used in their treatment.

**Conclusion:** The difficulty of reaching a clinical diagnosis of certainty is reflected in the negativity of cultures in all three patients although all had a history of contact lens use. For this reason they had received combined antibacterial and ant-amoebic treatment and one of them antifungal.

#### PS-11-013

##### **New diagnostic approaches to uveal melanoma: EGFR, TGFb and MMP9 expression correlate with histological type and invasiveness**

N. Danilova\*, S. Davidova, P. Malkov

\*Lomonosov Moscow State University, Dept. of Pathology, Russia

**Objective:** The purpose of the present study was to investigate the relationship between MMP9 expression and scleral invasion of uveal melanoma (UM). We also examined the effect of growth factors (TGFb and EGF), oncosuppressor protein (p16) on the histological types and mitotic activity of tumour.

**Method:** Tumour specimens were obtained from 42 primary UM immediately after enucleation.

**Results:** Hyperexpression of MMP9 and EGFR were correlated with a high proportion of spindle cells in UM (Kruskal-Wallis test  $p < 0,05$  for each). Moreover, we have demonstrated the association between the level of EGFR, TGFb and MMP9 expression to the initial stage of tumour invasion (Spearman's test  $p < 0,05$ ). Moreover, there was a correlation between TGFb hyperexpression and mitotic activity (Spearman's test  $p = 0,059$ ). Furthermore, a low level of p16 expression in UM was proportional hyperexpression of TGFb.

**Conclusion:** EGFR and MMP9 are known to be used as targets for anti-cancer therapy. The results of our study are suggesting to develop newer approaches of UM treatment on the early stages of invasion in order to keep an affected eye as an organ. Thereupon, it was concluded about the key-role of abnormalities in TGFb-pathway that cause the down-regulation of p16-gene, where the latter may lead to increased mitotic rate.

Tuesday, 5 September 2017, 09:30–10:30, Hall 3

#### PS-12 Cardiovascular Pathology

#### PS-12-001

##### **Skeletal muscle biopsy in the diagnostic algorithm of rare cardiomyopathies - a retrospective study**

M.-C. Dinca\*, D.-A. Costache, P.-B. Harsan, C. G. Socoliuc, E. Gramada, E. Manole, A. Bastian

\*Colentina University Clinical Hospital, Dept. of Pathology, Bucharest, Romania

**Objective:** To highlight the usefulness of performing skeletal muscle biopsies in the work-up of cardiomyopathies with skeletal muscle involvement. Cardiac abnormalities are common during the evolution of many neuromuscular diseases, manifesting as dilated or hypertrophic cardiomyopathies, arrhythmias or conduction disturbances. While in most cases the onset is late, the cardiac symptoms may dominate the clinical picture or even precede the myopathic signs.

**Method:** We retrospectively reviewed 463 consecutive muscle biopsies performed and analyzed in a 3 years period (2014–2016), using the Colentina Clinical Hospital Pathology Department database.

**Results:** The skeletal muscle tissue was obtained using open biopsy under local anesthesia; muscle cryosections served for histological, histoenzimological, immunohistochemical stains and complementary techniques of western blotting and electron microscopy. We diagnosed two cases of desmin-related myofibrillar myopathies, one later proved to be caused by a newly identified form of mutation in desmin, the other carrying an additional mutation in alpha-B crystalline gene, two cases of acid maltase deficiency with cardiac involvement, one case of Danon disease, a multi-minicore myopathy with cardiomyopathy and one gamma sarcoglycanopathy with severe cardiac signs.

**Conclusion:** The morphological aspects enabled us to establish the diagnosis with major impact on clinical management and directed further confirmatory genetic testing.