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ORAL ABSTRACTS (AO)

MULTIPLE HETEROZYGOUS MUTATIONS IN CNNM4 CAUSE JALILI SYNDROME. *RENATO ASSIS MACHADO, CÉLIA MÁRCIA FERNANDES MAIA, VERA LÚCIA GIL DA SILVA LOPES, ELAINE LUSTOSA MENDES, HERCÍLIO MARTELLI-JÚNIOR, RICARDO D. COLETTA.*

Jalili syndrome (JS-OMIM #217080) is an autosomal recessive disease with ocular and dental manifestations, resulting in dystrophy of rods and cones in the retina and amelogenesis imperfecta. It is reported that JS is caused by mutations in locus 2q11, which encodes the metal transporter gene *CNNM4*. In the current report, we describe a 9-year-old girl with a diagnosis of congenital loss of vision (Leber's congenital amaurosis) and yellowish, brittle, and painless teeth (amelogenesis imperfecta). Genetic analysis revealed heterozygous missense mutations c.971T>C in exon 1 and c.1742C>G in exon 4 of *CNNM4*. In addition, a polymorphic variant rs41286594 in exon 5 and a noncoding mutation c.2319G>A in exon 6 were found. Our approach thus led to the identification of multiple heterozygous mutations in *CNNM4*, which contribute to the pathophysiology of JS. Acknowledgment: FAPEMIG and CNPq.

BOTRYOID RHABDOMYOSARCOMA: REPORT OF A CASE. *JOHN LENNON SILVA CUNHA, AMANDA FEITOZA DA SILVA, FRANCISCO DE ASSIS ALMEIDA LIMA-JÚNIOR, RAQUEL MACHADO OLIVEIRA, THIAGO DE SANTANA SANTOS, ERICKA JANINE DANTAS SILVEIRA, RICARDO LUIZ CAVALCANTI DE ALBUQUERQUE-JÚNIOR.*

Rhabdomyosarcoma is a rare oral soft tissue malignant tumor with pathologic features that may influence the clinical behavior, treatment, and prognosis of the lesion. We report the case of a 13-year-old female patient presenting with an asymptomatic polypoid swelling in the left jugal mucosa measuring approximately 2.5 cm in diameter and of 3 months evolution. The presumptive diagnosis was fibrous hyperplasia. An excisional biopsy was carried out. Pathologic analysis revealed proliferation of predominantly ovoid cells, with eosinophilic cytoplasm and pleomorphic nuclei, arranged in the subepithelial cambium layer. The mucosal surface presented a papillary–verrucous appearance. Immunohistochemical analysis revealed intense positivity for desmin, myogenin, and ki67. The diagnosis was embryonal rhabdomyosarcoma (botryoid variant). The patient underwent complementary chemotherapy and radiotherapy, with no evidence of recurrence or metastatic disease at the 4-month follow-up. The clinical, histopathologic, immunohistochemical, and therapeutic aspects of botryoid rhabdomyosarcoma will be discussed.

TYROSINE-RICH CRYSTALLOIDS IN PLEOMORPHIC ADENOMA: A RARE DESCRIPTIVE CASE. *GLEYSON KLEBER DO AMARAL-SILVA, AMANDA ALMEIDA LEITE, ALAN ROGER SANTOS-SILVA, FELIPE PAIVA FONSECA, ANDRÉ CAROLI ROCHA, OSLEI PAES DE ALMEIDA, PABLO AGUSTIN VARGAS.*

Pleomorphic adenoma (PA) is the most common salivary gland neoplasm and has several peculiar histopathologic features, such as tyrosine-rich crystalloids (TRCs), which can be present in 2% of PA cases. A 58-year-old woman underwent surgery to excise a parotid gland lesion. The surgical sample measured $30 \times 26 \times 24$ mm; hematoxylin and eosin slides implicated PA comprising neoplastic strands of epithelial–myoepithelial cells, with some duct-like structures scattered in abundant loose stroma with unusual basophilic flower-shaped structures. Millon's reaction, a histochemical staining procedure for tyrosine, and scanning electron microscopy/energy dispersive X-ray spectrometry (SEM/EDS) analyses were performed. Millon's reaction showed intense reactivity in these structures, and SEM/EDS features confirmed the presence of TRC. Clinical relevance and development of TRC in PA is still unclear because of the small number of reported cases. Taking this opportunity, we describe a case of TRC in PA and the related clinical management.

RHABDOMYOSARCOMA OF THE MANDIBLE INITIALLY DIAGNOSED AS DENTAL ABSCESS: A CASE REPORT. *ARIANE DE SOUZA OLIVEIRA, FERNANDA BORTOLOTTI, FRANCESCA MAIA FARIA, TATIANE CRISTINA FERRARI, LARA MARIA ALENCAR RAMOS INNOCENTINI, CRISTINA BUENO BRANDÃO, KARINA ALESSANDRA MICHELÃO GRECCA PIERONI.*

Rhabdomyosarcoma, accounting for 5-10% of childhood cancers and greater than 50% of pediatric soft tissue sarcomas, is a malignant tumor of skeletal muscle origin, common in the head and neck region. We report the case of a rhabdomyosarcoma, initially diagnosed as a dental abscess, in the mandible of a 7-year-old girl. Clinical examination revealed an expansive mass, 12 cm in diameter, in the body of the right jaw. On incisional biopsy, this was identified as a malignant neoplasm of low differentiation suggestive of sarcoma, requiring differential diagnosis between Ewing sarcoma and rhabdomyosarcoma. Expansion of the immunohistochemical panel showed positive results for desmin and HHF-35, favoring the diagnosis of rhabdomyosarcoma. The patient received neoadjuvant chemotherapy for cytoreduction. After complementary examinations, significant reduction of tumor mass was observed; thus, hemimandibulectomy could be performed. This case emphasizes the importance of accurate diagnosis of malignant neoplasms in childhood in view of sudden evolution of these diseases.

ECSTASY ABUSE AND ITS EFFECTS ON THE ORAL MUCOSA: TWO CASE REPORTS. *MARIEL RUIVO BIANCARDI, ROSE MARIA ORTEGA, DANIEL RICALDONI DE ALBUQUERQUE, VALFRIDO ANTONIO PEREIRA FILHO, LUCIANA YAMAMOTO DE ALMEIDA, JORGE ESQUICHE LEÓN, ANDREIA BUFALINO.*

Ecstasy (3,4-methylenedioxymethylamphetamine) is an illicit drug that has been increasingly abused by young people. Its oral manifestations include bruxism, increased incidence of caries, xerostomia, and oral ulcers. We report the cases of 2 young women (22- and 27-years-old) who presented with multifocal oral erosions and ulcerations. The lesions were painful and covered by a