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Abstracts

representing a biomarker for thrombopoiesis and neonatal clinical outcomes pathology.

E-PS-15-027

PD-L1 expression in peripheral neuroblastic tumours: the first evidence of a possible therapeutic target

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Background & objectives: Neuroblastoma is the most common among peripheral neuroblastic tumours and it represents the third most common childhood neoplasm. In the present study, we evaluated the PD-L1 expression by immunohistochemistry in peripheral neuroblastic tumours as a new possible therapeutic target.
Methods: We retrospectively collected 17 patients with peripheral neuroblastic tumours (15 poorly differentiated neuroblastomas, 1 differentiating neuroblastoma, 1 nodular ganglioneuroblastoma). Formalin-fixed-paraffin-embedded tumour samples were tested with PD-L1 SP263 assay. The tumour proportion score (TPS) was assessed, and the combined positive score (CPS) was calculated as well. A semiquantitative assessment for the extent of diffuse/fibrillary PD-L1 was also conducted.
Results: PD-L1 positivity was found in 7 cases (41%). Three cases showed PD-L1 membranous and cytoplasmic positivity both in tumour cells (2 with intermediate expression, 1 with only focal staining) and immune cells; the remaining 4 cases showed PD-L1 staining restricted to the immune cells. PD-L1 expression in the immune cells was diffuse (60%) in one case, intermediate (10-40%) in 5 cases and focal (2%) in the remaining case.
Although in this study only membranous PD-L1 staining was considered positive, other staining patterns were observed (e.g., PD-L1–positive fibrillary staining in the well differentiated, Schwannian stroma component in 4 neuroblastomas, and membranous staining restricted to the ganglion cells in one case).
Conclusion: Neuroblastoma is not generally a highly immunogenic tumour. However, in the present study, we have found a good percentage of these tumours with PD-L1 expression; this expression appears to be higher in better differentiated tumours, e.g. differentiating neuroblastoma and nodular ganglioneuroblastoma. This finding may represent a new potential therapeutic target for these tumours. However, due to the rarity of these paediatric neoplasms, more studies on larger series are needed.

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A mass that grows with the baby: a case of fibrous hamartoma of infancy

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Background & objectives: Fibrous hamartoma of infancy (FHI) is a benign rare neoplasm affecting infants. FHI is characterized by triphasic morphology consisting of fibroblastic fascicles, nodules of primitive cells and mature adipose tissue. In this case, we report FHI in a 9-months-old girl.
Methods: A lumbosacral mass specimen, measuring in total of 7x4x3 cm, on cut surface with a tan-yellow color consisting of haemorrhagic, fibrotic and lipomatous areas was sampled. Paraffin-embedded tissue sections of this specimen were evaluated for the histopathological characteristics. Demographic, clinical and imaging data were collected.
Results: A 9-months-old girl admitted to the hospital with complaint of a congenital lumbosacral mass which was initially thought to be multiple Mongolian blue spots. Mother described the mass as it had been growing as the baby grew, but the growth rate has increased in the last few months. Her examination showed a moveable, painless mass with bluish color at the lumbosacral region. Magnetic resonance imaging revealed a subcutaneous mass with 5,5 cm in greatest width showing contrast enhancement. Histopathological examination revealed a characteristic organoid triphasic morphology with bland fibroblastic fascicles, mature adipose tissue and primitive looking oval-to-stellate shape cell nodules with myxoid stroma. Diagnosis of FHI was made.
Conclusion: FHI is a rare benign soft tissue neoplasm affecting mostly infants under 2 years of age. Rapidly growing nature and difficulties in making diagnosis may lead to misdiagnosis of this entity. While excision is curative, it is known that local recurrences could be encountered. Also, recent studies have shown that FHI cases often harbor EGFR exon 20 insertion/duplication mutations. Therefore, FHI should be considered in the differential diagnosis of a rapidly growing soft tissue mass in an infant.

E-PS-15-029

Paediatric melanoma: about two observations

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Background & objectives: Paediatric melanoma is rare. It represents for 3% of all paediatric cancers. Our objective is to report two observations of post pubertal paediatric melanomas.
Methods: Our observations concern two adolescents: Young girl of 14 years of clear phototype, followed for treatment of a pigmented lesion of the knee which appeared 04 years ago. A 17-year-old boy followed for axillary metastasis of melanoma of unknown primary. No personal or family history of nevus or melanoma was found in our two patients.
Results: We have examined:
- in the 14-year-old girl: a skin lesion measuring 34x20x12 mm, polypoid, ulcerated, infiltrating the hypodermis, poorly pigmented, strongly expressing HMB45, Melan A and Sox10. Ki67 was above 80%. Testing for the BRAFV600E mutation by RT-PCR came back negative. We retained the diagnosis of nodular post-pubertal melanoma.
- in 17-year-old boy: a lymph node massively infiltrated by a proliferation of poorly pigmented epithelioid cells, strongly expressing HMB45 and Melan A with mutation of BRAF V600E gene by RT-PCR. All clinical, endoscopic and radiological explorations did not find the primary melanoma.We retained the diagnosis of metastasis melanoma with unkown origin.
Conclusion: The particularity of our two observations lies in the development of melanoma in adolescents without a pre-existing naevus (dysplastic naevus, congenital naevus, Spitz naevus). Also, we found ourselves with an even rarer and more serious form of lymph node metastasis with no known primary melanoma in a 17-year-old adolescent.

The rarity of paediatric melanoma requires caution before confirming this diagnosis.

E-PS-15-030

Congenital intracranial adamantinomatous craniopharyngioma: a case report with histopathological and molecular characterization

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Background & objectives: Congenital intracranial tumours are exceedingly rare neoplasms that only account for 1% of brain tumours in childhood. To our knowledge, only thirty congenital intracranial