

CONGENITAL FIBROUS TUMOUR OF THE NECK WITH CHROMOSOME 11 MONOSOMY

Laine VJO, M.D., Ph.D.

Department of Pathology, Turku University Hospital, Turku, Finland

INTRODUCTION: Infantile (congenital) fibrosarcoma is a malignant childhood tumour with significantly better prognosis than fibrosarcoma of the elderly. It is a painless soft tissue mass of distal extremities or head and neck regions composed of small immature fibroblasts and high vascularity. Typical chromosomal abnormality in infantile fibrosarcoma is translocation t(12;15)(p13;q25) with a fusion of ETV6 and NTRK3 genes. Occasional findings are trisomies 8, 11, 17, and 20.

CASE REPORT: Newborn girl had a large congenital tumour in the neck resembling fibrosarcoma, with typical histology (small immature fibroblasts and haemangiopericytomatous vascular pattern). In MRI the tumour showed high vascularity, resembling hemangiopericytoma or congenital fibrosarcoma. The tumour was strongly immunopositive with vimentin and CD56 and weakly positive with smooth muscle actin. S-100 and neuroendocrine markers were negative as well as the fluorescence in situ hybridisation for the translocations t(X;18)(p11;q11) and t(11;15)(p13;q25), typical of synovial sarcoma and infantile fibrosarcoma, respectively. Translocation analyses were negative, but 20 % of the tumour cells showed chromosome 11 monosomy (typical of paraganglioma). The tumour was resected in part and the patient was treated with chemotherapy. The majority of the tumour could be resected and the patient is alive and well after two years' follow up.

CONCLUSION: We present here the tumor resembling congenital fibrosarcoma, but with unusual immunopositivity with CD56 and with chromosome 11 monosomy. Whether this is a new subtype of congenital fibrosarcoma or a paraganglioma with very unusual spindle cell histology remains obscure.