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Immunolocalization and Expression of Afadin-6 in Plexiform Neurofibromas

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Background
Neurofibromatosis type 1 (NF1) is a frequent single-gene disorder that affects the musculoskeletal system and the nervous system. Neurofibromatosis type-1 is inherited in an autosomal dominant manner with an incidence of about 1 in 3000. Tight junctions are specialized cell-cell point of adhesion at the apical region of epithelial and endothelial cells that creates cellular barrier. The Afadin-6 protein is a protein that contains two potential Ras binding domains. The Afadin-6 functionally links the cytoskeleton, through cellular signalling pathways and the cell-cell junctions. This study was carried out to demonstrate the relative expression and cellular localization of Afadin-6 in Plexiform neurofibroma by immunohistochemistry.
Methods
Informed patient consent was obtained two weeks before surgery and the study has an ethical approval (06/1505/137) of Liverpool Research Ethics Committee. Standard Operating Procedure of the department of Pathology, University of Liverpool was used in the immunohistochemistry technique. Both the test and control tissues were immunostained with Rabbit Anti-AF-6 polyclonal antibody diluted at 1:100-1:200 at (pH 7.0) (Catalogue No. 433280, Invitrogen). Slides were visualised under light microscopy.
Results
The Afadin-6 immunoreactivity on the perineurial fibroblast cell-cell junction was observed to be weak and localised at the cell-cell junction of the perineurial fibroblast of all the familial. Furthermore, moderate membranous and nuclei immunolocalization of the AF-6 were observed in endothelial and Schwann cells of all the Plexiform Neurofibromas.
Conclusion
The study suggests that Afadin-6 may be involved in cell proliferation and survival of the neurofibroma cells and therefore becomes a target protein in the management of plexiform neurofibroma which has the potential of transforming to Malignant Peripheral Nerve Sheath Tumour.

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