GENETICS AND GENODERMATOSES

UNILATERAL SEGMENTAL NEUROFIBROMATOSIS – A RARE PRESENTATION OF A COMMON GENODERMATOSES.

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BACKGROUND: Segmental neurofibromatosis(NF) or Type V neurofibromatosis is a rare type of genodermatoses in which café-au-lait macules and/or neurofibromas are limited to one region of the body. This occurs due to mosaicism resulting from somatic mutations. Early somatic mutations cause generalized disease that is clinically similar to the non-mosaic forms. Somatic mutation at a later point gives rise to segmental forms. Segmental NF is characterized by disease features limited to a localized area, which varies from a narrow strip to one quadrant and occasionally to one half of the body.

OBSERVATION: A 14-yr-old female presented to us with a history of nodular lesions on right side of face since two years' and pigmented lesions over right half of body since she was 1 year of age. She had congenital pseudoarthrosis of right tibia and fibula with limb length discrepancy since birth. On clinical examination, she had café-au-lait spots varying in size from 0.5cm by 0.5cm to 7cm by 5cm in size localized to the right half of her body (not crossing the midline) and flesh coloured, dome-shaped, soft to firm nodules on right cheek with facial asymmetry. B/L axillary and palmar freckling was present. Ophthalmic evaluation was in normal limits and she had no other comorbidities. Neurofibromas, café-au-lait macules and skeletal abnormalities were limited to the right side of the body.

KEY MESSAGE: Not many cases of Segmental NF have been reported so far. Our case is all the more unique as its presentation is limited to the right half of the body and not crossing the midline. To the best of our knowledge there haven't been any cases reported with a unilateral presentation. Mosaic forms of genetic diseases are known to have a lesser severe course and a lesser recurrence risk in offspring as compared to non-mosaic forms.