

GENETICS AND GENODERMATOSES

## SCOPE OF ULTRASTRUCTURAL STUDIES IN UNDERSTANDING LESIONS OF NEUROFIBROMATOSIS USING ELECTRON MICROSCOPY

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Introduction: Genodermatoses are a group of inherited disorders encompassing a broad range of inheritable skin diseases that can be associated with mortality and long-term morbidity. An accurate diagnosis is essential in circumventing associated systemic complications and advising against reproduction in some cases. Ultra-structural investigations can provide critical information regarding skin changes associated with diseases that otherwise rely largely on the physicians clinical acumen and have very few definitive diagnostic modalities.

Objective: Skin from neurofibroma (NF), Café-au-lait macule (CALM) and non lesional skin from the same patient are to be compared to assess:

- 1) Electron micrographs of epidermis
- 2) Electron micrographs of collagen in the dermis
- 3) Quantification of melanosomes
- 4) Qualitative analysis of melanosomes

Materials and Methods: Biopsies of following lesions (as were present) were taken from each of the 15 patients:

- 1) Non lesional skin
- 2) Café-au-lait macule (CALM)
- 3) Neurofibroma (NF)

Following investigations were performed on all three samples:

- 1) Histology of affected tissue using Hematoxyling & Eosin stains
- 2) Histology of affected tissue using Masson Fontana stain (for melanin)
- 3) Ultrastructural study using imaging tool- Transmission electron microscope

Results: Ultrastructural studies revealed:

1) An increase in melanin content in the epidermis of NF (NF>CALM> Non-lesional skin)











2) Number of melanosomes maximum in lesions of NF (>3 times that in non lesional skin), followed by CALM

3) Dense collagenisation observed in the dermis of NF compared to CALM and non-lesional skin

- 4) A type 2 macromelanosome in keratinocyte measuring 780nm-960nm was observed
- 5) Granular macromelanosome was spotted in the epidermis
- 6) Autophagosome-like vesicles were seen in epidermal cells

Conclusion: Electron microscopy based ultrastructural studies can increase our understanding of the disease process and provided a basis for interpretation of biochemical and physiological studies, at the same time proving as a valuable tool in diagnostics.





