

Phakomatoses refers to a group of neuro-oculo-cutaneous syndromes or neurocutaneous disorders involving structures arising from the embryonic ectoderm. These multisystem disorders involve the ectodermal structures like central nervous system, skin and eyes.

The lesions have a variable severity.

However, it has been subsequently noted that mesodermal and endodermal tissues too are involved.

A number of genetic and acquired diseases come in this category and may affect one or more of these tissues. However, in some conditions, such as von Hippel-Lindau disease, ectodermal presentation is minimal.

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Last update: **2024/06/07 02:52**

