Phakomatoses are a group of neurocutaneous disorders characterised by involvement of structures that arise from the embryonic ectoderm (thus central nervous system, skin and eyes). Other organs may also be involved.

As a group they are characterised by often widespread abnormalities often with characteristic appearances. Examples of phakomatoses are listed here:

neurofibromatosis neurofibromatosis type 1 (NF1) (von Recklinghausen disease)[+] neurofibromatosis type 2 (NF2) (mnemonic) tuberous sclerosis (Bourneville-Pringle disease) tuberous sclerosis diagnostic criteria ataxia telangiectasia Sturge-Weber syndrome (encephalotrigeminal angiomatosis) von Hippel-Lindau disease (retinocerebellar angiomatosis) incontinentia pigmenti (Bloch-Sulzberger syndrome) basal cell naevus syndrome (Gorlin-Goltz syndrome) Wyburn-Mason syndrome (Bonnet-Dechaume-Blanc syndrome) encephalocraniocutaneous lipomatosis hypomelanosis of Ito Nijmegen breakage syndrome epidermal naevus syndrome Schimmelpenning syndrome phacomatosis pigmentokeratotica nevus comedonicus syndrome angora hair nevus syndrome Becker nevus syndrome Proteus syndrome type 2 segmental Cowden disease CHILD syndrome FGFR3 epidermal nevus syndrome

Neurocutaneous melanosis

progressive facial hemiatrophy (Parry-Romberg syndrome) PHACE syndrome Cowden disease Gomez-Lopez-Hernandez syndrome