

Clinical Manifestations

Learning Deficits/Cognitive Disorders

- Attention deficit – 60%
- Learning disabilities - 50-75%
- Motor deficits
- Spatial deficits
- Autism spectrum disorders – 30%

Nervous System Disorders

- Neurofibromas – 50%
- Seizures – 10%
- Headaches – 20%

Visual Impairments

- Lisch nodules on the iris – 90%
- Retinal hamartomas ("small percent")
- Optic gliomas – 15%

Malignancies

- Malignant peripheral nerve sheath tumors – 3%
- Pheochromocytoma – 1%
- Chronic myeloid leukemia ("rare") brain tumors – 3-5%

Vascular Disease

- Dysplasia of blood vessels – 1%
- Hypertension ("frequent")

Musculoskeletal Disorders

- Focal scoliosis and/or kyphosis of the spine – 10-25%
- Sphenoid bone dysplasia – 5-10%
- Congenital hydrocephalus – 1-5%

Skin Conditions

- Café-au-lait spots – 95%
- Dermal neurofibroma – 95%

