

Blue rubber bleb nevus syndrome (BRBNS) is a rare blood vessel (vascular) disorder that affects the skin and internal organs of the body. Multiple distinctive skin lesions are usually characteristic of this disorder and are often present at birth or present during early childhood. Lesions in the gastrointestinal tract frequently become apparent during childhood or early adulthood. The lesions are multifocal venous malformations, resulting from abnormal embryonic blood vessel development. BRBNS is characterized by soft, elevated lesions on the skin or just under the skin that are dark blue, red, purple-red or black in color. The venous malformations may be tender, contain blood and be easily compressed and are usually located on the upper limbs, trunk and soles of the feet but can occur anywhere. The lesions increase in size and become more apparent over time but have not been reported to become cancerous. The organ system most commonly affected by BRBNS is the gastrointestinal (GI) tract, particularly the small intestine. The lesions in the GI tract often bleed and can lead to mild or severe anemia. Iron replacement and/or frequent blood transfusions may be required. The GI lesions can also cause an obstruction or blockage (intussusception) of part of the bowel. Skeletal abnormalities and venous malformations in muscle are sometimes associated with BRBNS. Blue rubber bleb nevus syndrome is sporadic. Although families have been described in which the condition follows autosomal dominant inheritance, these families actually have other multifocal venous malformations. Blue Rubber Bleb Nevus Syndrome affects males and females in equal numbers. Approximately 150 cases have been reported in the medical literature. BRBNS is diagnosed by physical examination and a procedure in which the GI tract is illuminated and visualized (endoscopy). Genetic testing for BRBNS is available on a research basis only.