Skeletal Atavism UCDavis CA USA

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Skeletal atavism is a heritable defect of bone development that occurs in Shetland ponies and American Miniature horses. It is characterized by the ulna (bone next to radius) and fibula (bone next to tibia) growing too long, failing to fuse to the radius and tibia and leading to abnormal limb development. The defect affects both front and hind limbs, resulting in severe angle anomalies and deformation of the front knee and hock. Horses with skeletal atavism typically display short legs, a low rectangular shape body, narrowing at the knees, clubfoot and impaired movement. The angles of the legs and pattern of movement progressively worsen as the foal ages, and in most cases, the horse has to be euthanized within six months. A team of researchers in Sweden identified two independent, overlapping regions in the SHOX gene where DNA sequences were lost (deletions) in affected ponies. The deletions (**Del1 and Del2**) are of different sizes with the larger deletion (Del1) being more common among ponies.

Skeletal atavism is inherited as a simple recessive trait. It is estimated that about 12% of Shetland ponies are carriers. Regardless of sex, horses that carry two copies of the defective gene, either two copies of Del1, two copies of Del2, or one of each, are affected. A genetic test has been developed for skeletal atavism to assist owners and breeders to identify carriers and to make informed choices of mating pairs to avoid producing affected foals. Matings between carriers can result in 25% of foals having skeletal atavism and should be avoided.

The VGL is licensed to perform the genetic test for skeletal atavism.

ORDER TEST | PRICE LIST
Allow 6 -10 business days for results.

Results are reported as:

N/N	Normal. No copies of the skeletal atavism mutations detected.
N/Del1	Carrier. 1 copy of a skeletal atavism mutation detected.
N/Del2	Carrier. 1 copy of a skeletal atavism mutation
Del1/Del1	Affected. 2 copies of mutation detected.
Del2/Del2	Affected. 2 copies of mutation detected.
Del1/Del2	Affected. 2 copies of mutation detected.

More information about skeletal atavism is available at http://www.capiletgenetics.com/en/skeletal-atavism-test-eng

Reference:
Rafati N, Andersson LS, Mikko S, Feng C, RaudseppT, Pettersson J, Janecka J, Wattle O, Ameur A, Thyreen G, Eberth J, Huddleston J, Malig M, Bailey E, Eichler EE, Dalin G, Chowdary B, Andersson L, Lindgren G, Rubin CJ. "Large Deletions at the SHOX Locus in the Pseudoautosomal Region Are Associated with Skeletal Atavism in Shetland Ponies." *G3: Genes|Genomes|Genetics* (Bethesda), 6(7):2213-2223 (2016).