

Pituitary dwarfism in German shepherd dogs and Saarloos wolfhounds **-Availability of a genetic test-**

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The pituitary is a hormone producing gland at the base of the brain. This pea-sized gland is composed of three parts: the anterior pituitary, the intermediate lobe and the posterior pituitary. The anterior pituitary synthesizes and secretes six hormones, which are essential for numerous body functions, such as growth, reproduction, lactation, metabolism and handling stress. These 6 hormones are:

- Growth hormone (GH), which is essential for growth
- Thyroid stimulating hormone (TSH), which regulates thyroid function
- Prolactin (PRL), which is essential for lactation
- Follicle stimulating hormone (FSH) and Luteinizing hormone (LH), which are essential for ovulation in female animals and sperm production in male animals
- Adrenocorticotroph hormone (ACTH), which stimulates the adrenal cortex

Any defect in the development of the pituitary gland may result in a form of isolated or combined pituitary hormone deficiency. In dogs, congenital GH deficiency or pituitary dwarfism is the most striking example of pituitary hormone deficiency. This recessive inherited disorder is encountered most often in German shepherd dogs, but it has, for example, also been reported in Saarloos wolfhounds. The genetic defect causing congenital GH deficiency in German shepherd dogs is also the cause of pituitary dwarfism in Saarloos wolfhounds, because the disorder in the latter breeds was first recognized after German shepherd dogs had been used in the breeding of Saarloos wolfhounds.

German shepherd and Saarloos wolfhound dwarfs have a combined deficiency of GH, TSH, PRL, and the gonadotropins. In contrast, ACTH secretion is preserved in these animals.

Dogs that are carriers of the mutated gene that causes pituitary dwarfism do not have any symptoms and look exactly the same as the dogs that aren't carriers. Since pituitary dwarfism is a recessive disorder of a single gene, the birth of a dwarf indicates that both parents are carriers of the mutation.

Pituitary dwarfs are significantly smaller than their healthy littermate, but the dwarfs are in proportion. Another clinical manifestation of pituitary dwarfism is that the dwarfs have retained their puppy hair coat. In time, the hair coat will be largely lost and the animal will become alopecic (bald) (Figure 1).

The growth retardation and the abnormal hair coat are mostly noticed by 2 to 3 months of age. The hairs are easily epilated, and when the animal loses its hair coat, the skin can become squameous and hyperpigmented, making the skin darker in color. Furthermore, due to a lowered local immunity of the skin, dwarfs are prone to bacterial skin infections.

However, the clinical signs are not limited to exterior appearances. The dwarfs suffer from a whole range of clinical manifestations far worse than skin and hair coat problems. For instance, GH deficiency also leads to underdevelopment of the kidneys, causing chronic renal failure. The deficiency of TSH will result in an underactive thyroid gland, causing the animals to be slow and dull. Furthermore, the insufficiency of the gonadotropins will result in failure of one or both testis to move, or "descend" into the scrotum (cryptorchidism) in male dwarfs. Female dwarfs do go into heat, but they do not ovulate. It can be concluded that pituitary dwarfism is a serious disorder.

Although the physical features of pituitary dwarfism may seem obvious, the final diagnosis should be based on 'pituitary stimulation tests'. These tests can detect a deficiency of GH, TSH, prolactin, LH and FSH.

The most logical therapeutic option would be to treat the dwarfs with canine GH and thyroid hormone. Treating the animal with thyroid hormone is simple, but it is not possible to treat the dwarfs with canine GH, since it is not available for therapeutic use. However, research has demonstrated that porcine GH is identical to canine GH, making it a good alternative for treatment.

Without proper treatment, the long term prognosis is poor. Many dwarfs will not live more than 4 to 5 years. However, some dogs do live longer, probably because in some cases the pituitary still produces a small amount of hormones. Although the prognosis improves significantly when dwarfs are properly treated, their prognosis still remains guarded.

It should be clear that the birth of dogs with this serious illness should be prevented. In order to do so, two carriers of this mutation should not be bred. The problem is that, as mentioned earlier, one cannot distinguish a carrier from a non carrier judged on its appearance. This would require a genetic test. After 15 years of intensive research at the Department of Clinical Sciences of Companion Animals of Utrecht University, this test is now available! If this test would be used for all breeding animals, pituitary dwarfism could be completely eradicated in German shepherd dogs and the Saarloos wolfhounds.

A genetic test may not seem to be of big importance, since the disorder seems to occur only occasionally. However, one should keep in mind that many dwarfs die in the uterus or shortly after birth. One should also be aware of the fact that if just 1 percent of the dog populations are dwarfs, then 18 percent of the population will be carriers of the mutation. This means that the number of carriers will be much higher than might be expected. When 2 of these carriers are mated, on average 25 percent of their offspring will be dwarfs and half of the siblings will be carriers of the mutation.

For the genetic test, 4 ml. of blood (collected in an EDTA containing tube) is needed. The blood sample has to be shipped to:

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The costs of the genetic screening for the mutation that causes pituitary dwarfism in German shepherd dogs and Saarloos wolfhounds are Euro 100 (excl. VAT).

In short, pituitary dwarfism is a serious, incurable illness of which the occurrence is highly underestimated! The good news is that there is now a genetic test with which carriers of the mutation can be identified. If all breeding animals were tested (only once), and a correct breeding policy would be implemented, this severe illness could be completely eradicated.



Figure 1. A 10-month-old German shepherd dwarf with lack of guard hairs and retention of puppy hairs.