Clitoral Hypertrophy in a Weimaraner

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A 16-month old Weimaraner presented to AUCVM for evaluation of clitoral enlargement and ovariohysterectomy. Upon examination, the bitch was a phenotypic female with a severely enlarged clitoris, which initiated the investigation of a possible reproductive anomaly in this patient. Palpation of the clitoris revealed an os clitoridis and resulted in clitoral engorgement and pelvic thrusting. Pre-surgical blood samples were obtained for hormonal analysis and karyotype, and radiographs were taken to confirm the os clitoridis. During surgery, a uterus was exteriorized and gonads were located in a normal anatomical position consistent with ovaries. Interestingly, neither gonad was contained within an ovarian bursa. Grossly, the right gonad resembled a testis with an attached tubular structure resembling an epididymis; the left gonad more closely resembled an ovary. The uterus and both gonads were removed and submitted for histopathologic examination. The clitoris was surgically removed for cosmetic purposes.

Histologically, the right gonad consisted of numerous seminiferous tubules lined by Sertoli cells, and the interstitial tissue contained Leydig cells. Multiple cross sections of thin walled veins and arterial wall were visualized, suggestive of a pampiniform plexus. The left gonad had identical histologic morphology, but lacked the pampiniform like structure. The uterine horns and body were consistent with normal uterine tissue lined by endometrium.

Pre-surgical testosterone levels of 194.7 pg/ml were consistent with the presence of functional testicular tissue and comparable to that of a cryptorchid male (100-1200 pg/ml). Karyotype was determined to be 78XX and this patient was Sry negative. Normal female karyotype, presence of functional testicular tissue, and lack of concurrent ovarian tissue led to a diagnosis of XX sex reversal. An abnormality of sexual development, XX sex reversal is seen in American Cocker Spaniels and has been reported in 18 other breeds, including the Weimaraner. Investigations into the genetic mechanism leading to Sry negative XX sex reversal have resulted in the exclusion of several possible candidate genes. Recent research suggests a novel locus on CFA29 may be responsible for the condition seen in American Cocker Spaniels; however the exact gene or genes responsible for XX sex reversal remain unknown. This case demonstrates the use of diagnostic testing to determine the cause of reproductive anomalies.

Keywords: Sex reversal, genetic defect, clitoris, karyotype

References