



The Female Elite Athlete: Is it Genetic? A Review of Key Literature

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Introduction

For elite female gymnasts, a bias toward a prepubescent physique in combination with long training hours often results in delayed pubertal development. However, recent studies have suggested that delayed menarche may be caused by genetic factors and that girls who mature later often self-select sports that require relative strength and small bodies; such as gymnastics. This project was designed to investigate the effects of high intensity physical exercise on the hormonal regulation of puberty and to establish if a common gene, or genetic mutation, could be responsible for late maturation. Due to Covid-19, this project was restricted to a literature review.

The Female Triad

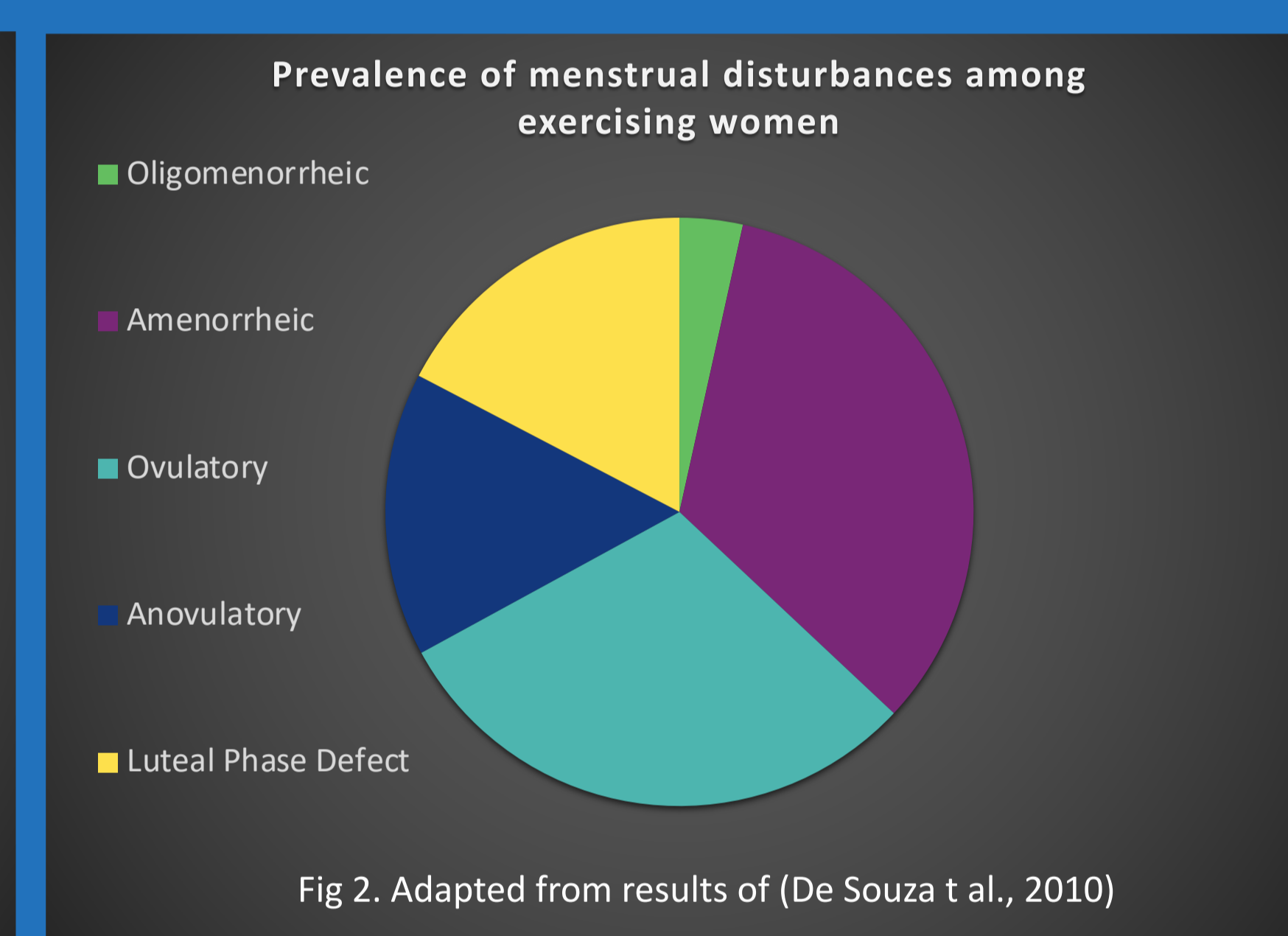
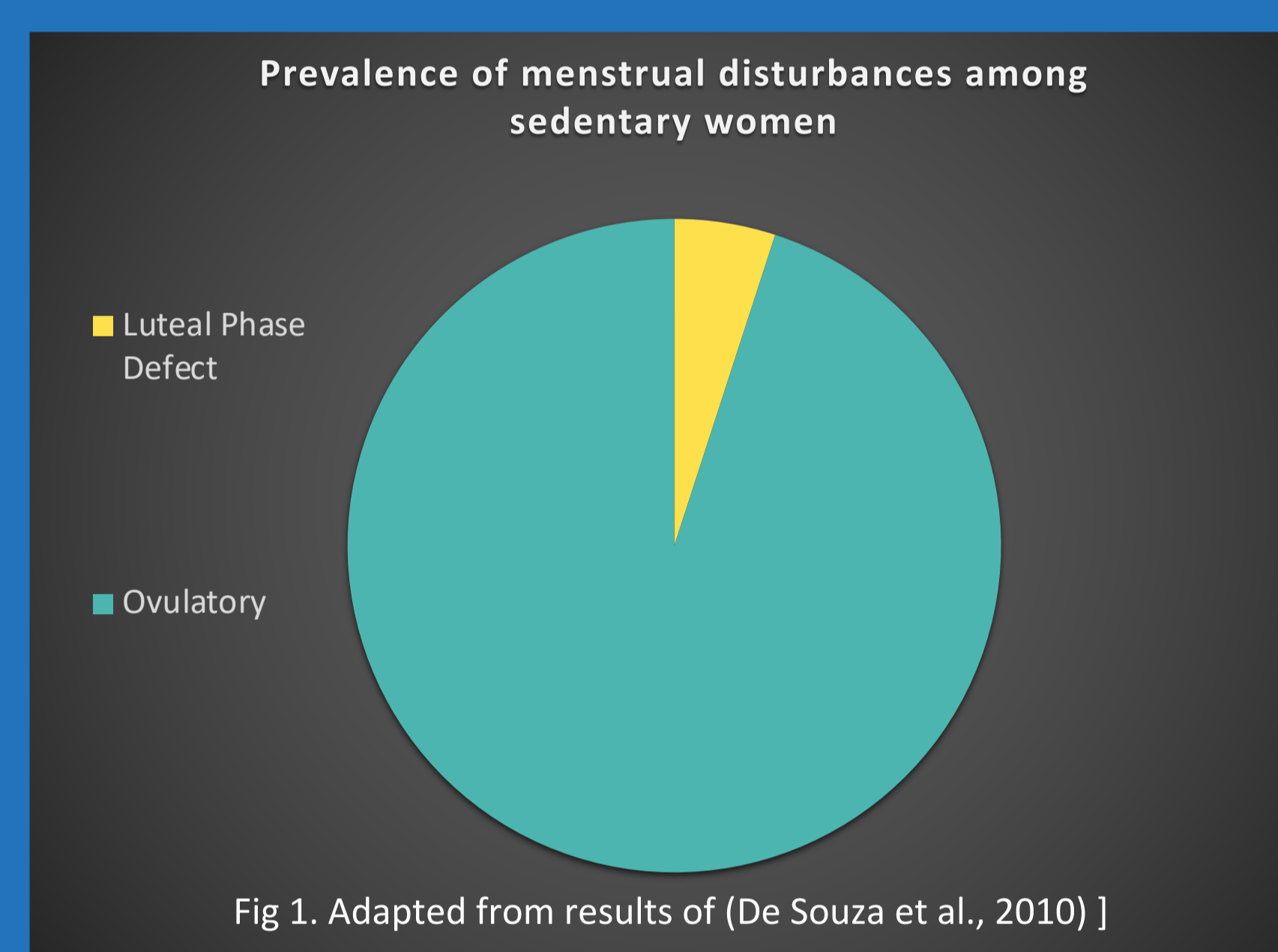
The three components of the female athlete triad are disordered eating, menstrual dysregulation and osteoporosis. It is becoming a more prominent and accepted theory with the increased interest in aesthetic and endurance sports that encourage high volume training at younger ages and lower bodyweights. Diagnosis of this is complicated and often requires specialists from several disciplines. The prevalence of clinical eating disorders among female elite athletes has been reported from 16% to 47% (1). Many female athletes may have disordered eating by inadvertently failing to meet the energy requirements to meet their large energy expenditures due to lack of nutritional knowledge (2). Many studies of anorexia nervosa report significant increases in total body fat percentage before the return of menses (3). This link to body fat percentage is frequently discussed among studies regarding female athletes, however causality has not been shown.

Hormonal Regulation

The exact genetic control of puberty remains a largely unanswered question. Many genome-wide association studies have observed that the timing of pubertal onset is likely to involve hundreds of genetic variants. However, menstrual disorders can derive from defects at any level of the hypothalamic-pituitary gonadal (HPG axis) and from neurological signals to the hypothalamus. In gymnasts with disruptions in their leutinising hormone (LH) cyclicity, as well as low oestrogen levels, their pituitary response to gonadotropin-releasing hormone (GnRH) provides confirmation that their disorders are secondary to the disruption of the hypothalamic GnRH pulse generator (4). However, the influence of athletic training on the GnRH generator is yet to be elucidated. A negative energy balance, which may result from reduced caloric intake and increased caloric expenditure, has been associated with reduced serum insulin-like factor I and has been theorised to cause delayed menarche, as it is known that a sharp rise in insulin-like factor I is experienced during puberty (5). A plausible hypothesis is that highly intensive training disrupts hormone cyclicity patterns and this requires further detailed study. This hypothesis allows for the appearance of normal menstruation patterns in gymnasts post-retirement.

Menstrual Disturbances

The variety of menstrual disorders present in female athletes could reflect differing degrees of exposure to a disrupting factor or differences in the susceptibility of various women to disruption. Amenorrhoea is defined as the absence of menses by 16 years, or the absence of menses for three or more months in menarcheal women. Oligomenorrhoea is defined as having a menstrual cycle that is greater than 36 days. Menstrual irregularities have been compared between elite rhythmic gymnasts and a normal control group. 79% of the gymnast participants, with a mean age of 14.5 (+0.2) years, had not yet menstruated. The menarcheal gymnasts were found to be significantly taller and heavier and to have lower training frequency and duration than the premenarcheal gymnasts. Of those menarcheal gymnasts, 78% reported either oligomenorrhoea or amenorrhoea. (6) Furthermore, menarche occurs within a small range of skeletal ages and intensive gymnastics training has been linked to skeletal maturation delays, which worsen with increasing years of training (7). It is important to note that in several studies, menstrual dysregulation has been linked to negative effects on bone mineralisation, and thus contributes greatly to the female athlete triad. The graphs below are adapted from a study that compared menstrual disturbances in sedentary and active women. Disturbances were found to be far greater in active women, with oligomenorrhoea and amenorrhoea having a much higher prevalence (8). However the sample size (n=86) was not very large.



Conclusions

Despite the high prevalence of delayed puberty, menstrual disturbances and reduced bone density seen in high-level gymnasts, no major studies have demonstrated a causal relationship between these factors and training intensity. As short stature and late maturation are advantages in gymnastics, bias in the selection process may result in individuals who genetically select for these traits. It has also been reported that mothers and sisters of elite gymnasts show signs of delayed puberty (9), and identical twin studies suggest that genetic factors account for a greater proportion of exercise behaviours than environmental factors do (10). Cross-sectional studies cannot infer causality therefore, more longitudinal and experimental research must be done. Further research must also aim to control for selection bias and to differentiate between actual and perceived time of pubertal maturation. Studying delayed puberty must overcome the added complication of the fact that this phenotype represents the tail of a normally distributed trait within the population and therefore the genetic variants that cause delayed puberty may also be present in the general population (11).

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