

# Premature thelarche from phenotype to genotype

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Premature thelarche is a benign condition, which has been described as a "variant of puberty". Early breast development is especially prevalent during the first year of life, when the gonadal axis is usually active. Ultrasensitive bioassays have described higher estrogen levels in these girls compared to controls. Some cases of premature thelarche may also exhibit increased growth velocity and/or bone age, despite prepubertal gonadotropin secretion. These cases have been labeled as exaggerated thelarche and may represent an intermediate state between benign premature thelarche and precocious puberty. Several factors have been associated with the etiology of premature thelarche, such as endocrine disruptors, and genetic and nutritional factors. Recently, it has been shown that some girls with exaggerated or fluctuating thelarche may have an activating mutation in the CNAS gene, which codifies for a subunit of G stimulating protein (Gs?). We discuss the different phenotypes that may be a