How is delayed puberty in girls defined?
Puberty starts when the pituitary gland begins to produce two hormones, luteinizing hormone (called LH) and follicle-stimulating hormone (called FSH), which cause the ovaries to enlarge and begin producing estrogens. The growth spurt starts shortly after breasts begin to develop, and the first menstrual cycle begins about 2–3 years later. A girl who has not started to have breast development by the age of 13 is considered to be delayed.

What causes puberty in girls to be delayed?
Some girls with delayed puberty are simply late maturers, but once they start, puberty will progress normally. This is called constitutional delayed puberty and is more common in boys than girls. Often, this is something that is inherited from the parents, so it is more likely to occur if the mother started her periods after age 14 (the average is about 12 ½) or if the father was a “late bloomer.”

Decreased body fat is a major cause of pubertal delay in girls. It can be seen in girls who are very athletic, particularly in gymnasts, ballet dancers, and competitive swimmers. It can also be seen in girls with anorexia nervosa, who engage in extreme dieting or binging and purging, because they fear becoming too fat even when they are abnormally thin. Finally, it can be seen in a number of chronic illnesses in which body fat is often decreased.

Some girls with delayed puberty may have problems with their ovaries. The ovaries are either not developing properly or are being damaged. This is referred to as primary ovarian insufficiency. The major cause present at birth is Turner syndrome, in which all or part of one of the two X chromosomes is missing. Most girls with Turner syndrome are also extremely short for their age and may have certain distinctive physical features, such as webbing of the neck, a high-arched palate, or arms that bend outward when extended. However, in most cases, Turner syndrome is diagnosed well before age 13 because of short stature. The major acquired cause of ovarian insufficiency is damage to the ovaries as a result of radiation, usually to treat leukemia or certain other kinds of cancer. Occasionally, girls may have their ovaries damaged by the body’s immune system.

Finally, some girls fail to start puberty because of a lack of the pituitary hormones LH and FSH, also called gonadotropins. This can occur when there are other pituitary deficiencies as well, including growth hormone, or it can be an isolated finding (particularly in a girl who is delayed but not short).

How is delayed puberty in girls diagnosed?
The endocrinologist will order blood tests to measure levels of LH, FSH, and estradiol and, in some cases, other tests. Very high levels of LH and FSH will indicate that the ovaries are not working properly, and the pituitary is trying to stimulate them to work harder. If the cause of the ovarian insufficiency is not clear, a chromosome study or karyotype will be done to see if all or some cells are missing all or part of an X chromosome. If the LH, FSH, and estradiol are all low, the problem could be either decreased body fat (if one of the risk factors listed above is present) or a permanent deficiency of LH and FSH. Other tests may be ordered if deficiency of multiple pituitary hormones is suspected, and on occasions, a brain MRI may be helpful. A hand x-ray for a bone age is often done, which is typically delayed by 2 or more years, which means that there is still additional time to grow.

How is delayed puberty in girls treated?
In girls with constitutional delayed puberty, breast development will eventually start on its own. Giving estrogens for 4–6 months is sometimes used to help get things started sooner. For girls with delayed puberty and decreased body fat, sometimes eating more and gaining weight will help get puberty started. For girls with primary ovarian insufficiency or a permanent deficiency of gonadotropins, long-term estrogen replacement is needed and can be given either in the form of a daily tablet of estradiol or as a patch that needs to be applied to the skin twice a week. Doctors usually start on a low dose and often increase the dose about every 6 months. After 12–18 months, it is typical to start a second hormone called a progestin (for example, Provera) which will, after a few months, result in a period, usually within a day or two of stopping the progestin. You may ask your endocrinologist to discuss with you and your child what is known about your child’s potential for fertility.

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