A young woman concerned about small breast size

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An unusually tall young woman is worried that her breasts have failed to develop as expected.

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CASE SCENARIO

Karen is 18 years old and presents because she is concerned about the small size of her breasts. She says that her breasts started to develop in her first year at high school but then seemed to stop growing, although she started menstruating at the age of 13 years and has had regular periods ever since. On examination, it is noted that she is very tall (180 cm), has a body mass index of 23 kg/m^2 and normal pubic and axillary hair and a normal female perineum. Her breasts show minimal development, with budding equivalent to Tanner Stage 2 of puberty. Karen has no other health concerns and is fit and active.

What should be the approach to Karen's concern?

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COMMENTARY

Karen presents with bilateral mammary hypoplasia, as the breast bud (Tanner Stage 2) is the first stage of the clinical onset of puberty in the female: Tanner Stage 1 is pre-puberty and Tanner Stage 5 is full adult (Figure).[1,2] There is nothing to suggest regression of normal development, given the menstrual history. Some women's breast never reach Stage 5, or do so only after pregnancy and lactation.

It is important to know whether the staging of breast development was performed by the clinician, or whether this is self-report based on comparison with Tanner Stage line drawings. The accuracy of the latter is limited, particularly if there is hypoplasia. Palpation is needed to confirm the presence and dimensions of glandular breast tissue. Tanner Stage 2 comprises glandular tissue under the areola only.

Diagnosis

The history of menarche and regular menstrual cycles is the most important part of this young woman's history. Menarche confirms that there is a uterus present with an endometrium capable of being stimulated, a source of oestrogen to cause endometrial growth and a vagina. Presumably the ovaries are producing endogenous oestradiol as there is no suggestion that Karen is taking medication. She should thus have adequate levels of oestradiol, the most important hormone in initial growth of lactiferous ducts and adipose tissue. Progesterone stimulates the alveolar growth of the breast tissue. The regular menstrual cycle is also consistent with the absence of any major abnormality of the endocrine system, normal body weight and generally sound physical health. An absence of menstruation (primary amenorrhoea) would greatly expand the differential diagnosis and require hypothalamic–pituitary–gonadal disorders to be considered.

Karen is tall (above the 95th percentile for height). A family height history is needed to comment further. A mid-parental height at or above the 85th percentile would be expected in this

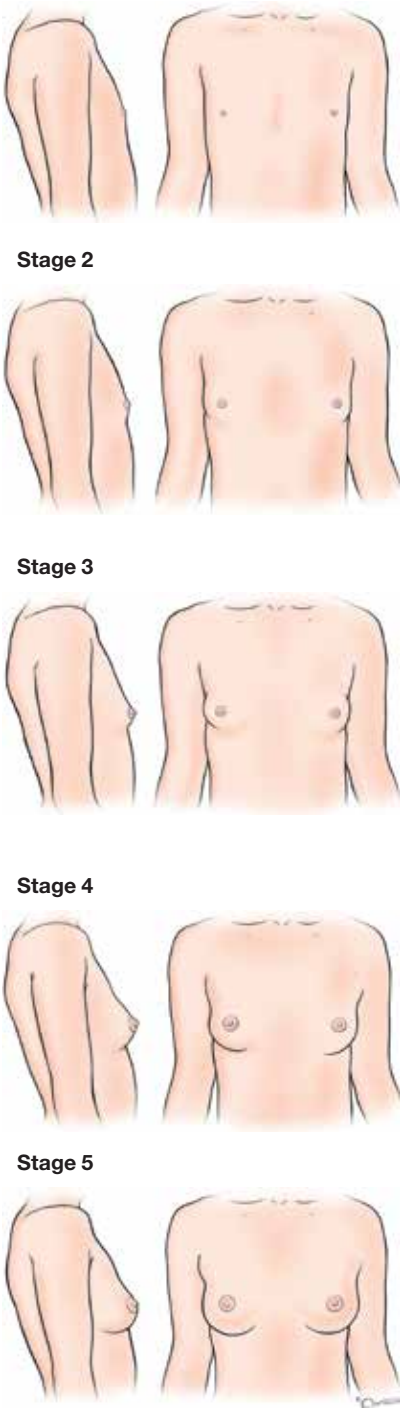

Figure. Tanner staging system for pubertal development of the female breast (Tanner, 1962).[2]

scenario. In the absence of a family history of tall stature, the differential diagnosis for Karen's height includes:

- idiopathic tall stature
- syndromal tall stature such as Marfan syndrome
- growth hormone excess secondary to a pituitary tumour prior to epiphyseal closure
- hypogonadal states with normal growth hormone dynamics, where growth continues because there is no epiphyseal fusion in the absence of gonadal steroids.

The final two possibilities are uncommon and are excluded clinically by Karen's general good health and normal menses. Syndromal tall stature might be considered because connective tissue disorders are associated with mammary hypoplasia. A history of joint hyperextensibility or dislocation might suggest a mild form of connective tissue disorder, but the therapeutic options are limited and would not address Karen's concerns about her breast size.

In summary, the adult female breast is highly variable in both shape and size. The amount of glandular breast tissue is modest in the nonparous woman. The hormones that contribute most to the development of the adolescent breast are oestradiol, progesterone, growth hormone, and circulating and local growth factors.

The most likely diagnosis in Karen's case is local under-responsiveness to endogenous hormones, leading to mammary hypoplasia.[3] If only one breast was involved then previous damage to a breast bud could be the cause, such as chest drain insertion as a neonate or the inexcusable biopsy/removal of a 'breast lump'. Poland syndrome, which includes the absence of the pectoral musculature, can also cause unilateral mammary hypoplasia.

Investigation and management

Karen should have biochemical confirmation that she has normal levels of oestradiol (sampled after the first seven days of the menstrual cycle to avoid the low levels seen in the perimenstrual period) and follicle-stimulating hormone to exclude early onset ovarian failure. This confirmation is to reassure the patient and to avoid unnecessary trials of exogenous oestrogen therapy. Although not routinely recommended, a breast ultrasound examination would define the amount of glandular tissue present and might provide reassurance about future breastfeeding capability, which is not related to breast size. Despite these suggestions and firm reassurance that there is no major pathology that requires treatment, Karen is still likely to be highly anxious and upset.

Management is cosmetic. There should be no expectation that the situation will improve with time. Karen has the choice of good quality prostheses worn as part of a bra, or cosmetic surgery. This situation and its resolution will be challenging for an 18-year-old. It will be necessary to explore issues around body image, and concerns about sexuality and reproduction. To do this, a clinician will require sensitivity and competent counselling skills. Most young women will ultimately choose surgery, but often not immediately. MT

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