Genetic selection and folate intake during pregnancy
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Serum leptin concentrations of 18 volunteers at 490 m (Lausanne, Switzerland) and at 4559 m (Capanna Margherita, Switzerland) with and without loss of appetite
An increase in integrated serum leptin levels (mean area under the curve) from 53.8 (SD 13.8) ng/ml per h to 66.3 (16.2) ng/ml per h was found in individuals with loss of appetite (1:20 h, p=0.008), but not in volunteers without loss of appetite (36.7 [6.4] ng/ml per h (490 m) vs 40.8 [13.2] ng/ml per h (4559 m), p=0.35).

with AMS, compared with concentrations at 490 m (AMS was defined as a functional Lake Louise Score of more than 1 [n=15, p=0.2]7). Statistics were Mann-Whitney test and Wilcoxon tests. Effects due to change in plasma volume were excluded. Individuals with loss of appetite showed a tendency to higher leptin concentrations at baseline than those without (p=0.1, figure), but mean body mass indices were not significantly different between subgroups.

In two independent studies, elevated leptin concentrations at high altitude were found to be associated with loss of appetite. Thus, leptin may be a player in the altered neuroendocrine regulation of energy homeostasis at high altitude, leading to loss of appetite, increased energy expenditure and weight loss.

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prevent neural-tube defects. This treatment was started by physicians prescribing multivitamins and folic acid some years earlier; 3% of pregnant women received multivitamins in 1976, which increased to 10% in 1977, 35% in 1982, and 55% in 1986. An association has been reported between women with the hyperhomocysteinemia and the A225V mutation and recurrent miscarriages and that these women have normal births after folic acid and pyridoxine supplementation. Therefore, a hypothesis can be made that there is an association between early folate supplementation during pregnancy and the increased number of babies born with the VV genotype, especially in VV mothers. The Mediterranean diet may also influence differences in genotype frequencies between northern and southern Europe.

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A 47,XXY female
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Klinefelter syndrome (47,XXY) with male phenotype is the most common sex chromosomal abnormality. It is believed that SRY (sex determining region on Y) is the major gene necessary to induce the undifferentiated bipotential gonadal primordium to develop as testis. Recently, several SRY-box-related (SOX) genes have been identified on autosomes. Mutations in the SRY or SOX gene have been implicated in sex reversal. We report an individual having 47,XXY chromosome constitution, normal SRY, SOX9, and ZFY genes and yet with a female phenotype.

A 15-year-old phenotypic female has well developed breasts; prominent labioscrotal folds; an enlarged clitoris; feminine pubic hair distribution; urogenital sinus 5 cm deep; a uterus; prepuce in shape and size, palpable per rectum; and a cervix which could not be palpated. Ultrasonography of the abdomen revealed normal shape of urinary bladder with smooth and regular walls. Uterus was small in size, with normal shape and outline. Myometrium was homogenous, cavity was empty, midline echo was normal. Laparoscopic assessment revealed a normal uterus. Right ovary was normal in size and shape with a cyst and a normal Fallopian tube. Left ovary was not seen as it was hidden beneath an ephephelial fold.

Chromosome analysis showed 47,XXX karyotype in all 100 metaphases analysed. Hormone profiles showed average female range of corticotrophin, T3, T4, thyrotrophin, and testosterone. Progesterone and prolactin were higher when compared with normal female levels. Histology of the right gonad showed features compatible with ovarian stroma. No germ cell, ovarian follicle, or corpora alibicans were detected in these areas. Left gonad showed occasional seminiferous tubule-like structures devoid of germ cells and spermatogenesis. Southern hybridisation and FISH with a Y-chromosome-specific probe established the presence of Y chromosome in blood and gonadal tissues and ruled out the possibility of mosaicism. PCR amplification and sequencing of the PCR products of the SRY, SOX9, and ZFY genes showed no mutation. X-chromosome specific STR analysis revealed the paternal origin of the extra X chromosome.

There are reports of mosaicism (46,XX/47,XXX and 46,XX/47,XXX/48,XXXXY) leading to true hermaphroditism. But in the present case mosaicism was ruled out. Generally, individuals possessing 47,XXX karyotype have male phenotype. However, in the present case in spite of the presence of Y chromosome and the normal SRY, SOX9, and ZFY genes, the individual has a female phenotype. This strongly suggests that the phenotypic sex in this case might be due to the involvement of other sex determining genes.

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Acute neuropathy after exposure to sun in a patient treated with St John’s Wort
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Extracts or whole-herb preparations of St John’s Wort (Hypericum perforatum) are popular to treat depression. Photosensitivity is known to be a possible side effect. We report one patient who, while taking St John’s Wort, developed subacute polynoephropath after exposure to sun.

A 35-year-old woman took ground whole St John’s Wort (500 mg/day) for mild depression, after reading a magazine article. After 4 weeks, she developed stinging pain on her face and dorsum of both hands (areas exposed to the sun). Spontaneous pain was mild but worsened during and after being in the sun. Pain was provoked by minimal mechanical stimuli such as light touch or air movement. Cooling increased and warming decreased the pain. She sought help when the same symptoms developed on her arms and legs a few hours after sunbathing, and were limited to the exposed skin.

She was examined 2 weeks after the pain started. There were no skin burns on examination. Light brushing, a gust of air at room temperature, and cold (~5-10°C) were acutely painful and outlasted the stimulus for seconds afterwards, consistent with alldynia. These symptoms were restricted to

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