LETTERS TO THE EDITOR

Osteoglyphonic dysplasia

We read with interest the manuscript "Osteoglyphonic dysplasia", by Professor Peter Beighton (J Med Genet 1989;26:572-6). However, we would like to voice an objection to the spelling of the name of this disorder. Professor Jürgen Spranger suggested the term 'osteoglyphonic dwarfism' on the basis of its radiographical findings. The metaphyses appear to be 'hollowed out', so the term 'osteoglyphonic' for 'hollowed bone' was proposed.

The Greek noun γλυφός (genitive γλυφόδος) refers to the notch of an arrow (by which it is seated on the bowstring) and, by extension, to the arrow itself. The related verb γλαφάω, γλαφάται means "to hollow out, engrave, carve", and, by extension, "to write" (on a tablet). The Greek root persists in English as the suffix "-glyph", "-glaryph" or "petroglyph". The same root appears in many western European languages as a word meaning "to cut" or "to cleave", for example, in cleft palate. The Greek letter upsilon should be translated into English only as y or u. There is no reasonable English equivalent which uses the letter o for the Greek upsilon.

Therefore, we feel that this condition should be named correctly either 'osteoglyphic' or 'osteoglyphicid' dysplasia.

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Choanal atresia as a feature of ectodactyly-ectodermal dysplasia-clefting (EEC) syndrome: a further case

Christodoulou et al reported a family in which the proband had choanal atresia associated with the EEC (ectodactyly-ectodermal dysplasia-clefting) syndrome. We wish to report another child with this syndrome, and choanal atresia, and confirm this feature to be associated with the EEC syndrome.

The proband presented with bilateral cleft of the lip and palate, preaxial polydactyly of the left foot, left hydropneophrosis, right dysplastic and non-functioning kidney, and web penis. His mother has bilateral cleft of the lip and palate with no nail, hand, foot, hair, or tear duct abnormalities. However, the sib has classical features of EEC syndrome, including choanal atresia. In addition to very fair, brittle hair, she has bilateral absence of the tear ducts, facial dysmorphism consisting of underdeveloped philtrum, flat nasal bridge, and lateral placement of the inner canthi, and, in addition, syndactyly of fingers 3 and 4 on both hands. She has vesicoureteric reflux.

The diagnosis of the EEC syndrome in the proband and mother would have been difficult without the affected sib. This family also indicates the extreme variability and the renal tract anomalies seen in this syndrome.

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Paraplegia and arthrogryposis multiplex of the lower extremities after intrauterine exposure to ergotamine

In a recent issue of this Journal, Hughes and Goldstein reported on a microcephalic girl with paraplegia, joint ankylosis, and anaesthesia of the lower limbs, suggesting medullar injury; lissencephaly and brain atrophy were also present. She was born after intrauterine exposure during the first four months of gestation to several vasoactive drugs: propranolol (80 mg/ day) and 'cafergot' suppositories (one to four/week).

We recently observed a child with arthrogryposis congenita and paraplegia, whose mother took ergotamine in the fourth month of pregnancy. Our proband, a girl, was born at 32 weeks of a dizygotic twin pregnancy, obtained through in vitro fertilisation. Her brother weighed 2300 g and was normal. Her weight was 1720 g (50th centile) and OFC was 31 cm. Arthrogryposis multiplex of the lower limbs with sensorimotor nerve defect was present. The symptoms were very similar to the neurological status of a spina bifida aperta (or any other spinal cord trauma involving segments L1 to S1): bilateral equinovarus deformity of the ankles, bilateral fixation of the knees at right angles, and bilateral luxation of the hips, which were fixed in abduction-internal rotation. Moreover, perpartal fractures of both femora were present. There was both faecal and urinary incontinence and anal eversion. The thighs and buttocks were hypoplastic. There was some spontaneous movement at the ankle joints, as well as hip flexion, and some sensitivity remained in the plantar area and around the hip. The upper limbs were not involved. The face was normal.

Subsequent psychomotor development was normal, and partial motor and sensory recovery was observed. Transfontanellar ultrasonography and EEG were normal. Spine x ray, CT scan, and NMR imaging of the lower medulla oblongata were normal and excluded extrinsic compression or vertebral malformation. Electromyography showed a denervation pattern of fibrillations with some bursts of voluntary contraction in the quadriceps muscles. Prenatal cord trauma seemed the most probable aetiology, considering the muscle atrophy and ankylosis.

The parents were normal, non-consanguineous Caucasians and there was no relevant family history. However, at 4½ months of gestation, the mother, who suffered from migraine, took one suppository of Cafergot® (Sandoz, composition: ergotamine 2 mg, caffeine 100 mg, belladonna alkaloid 0.25 mg, butalbital 100 mg). She suffered from severe side effects including intractable nausea, vertigo, and dizziness, which confined her to bed for three days. The rest of the pregnancy was uneventful. Neither hydramnios nor oligohydramnios was recorded, nor a decrease in fetal movements.

The vascular effect of therapeutic or toxic doses of ergot alkaloid has been widely documented in man. Individual sensitivity to therapeutic doses of
Choanal atresia as a feature of ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome: a further case.

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