Correspondence

prompts us to report a mother and daughter who presented with very similar features (fig 1).

The 4 year old daughter, who showed moderate developmental delay, was referred because of concern over her persisting large anterior fontanelle with wide metopic and sagittal sutures. Additional findings included facial asymmetry, prominent crura of both ears, and short digits with finger-like thumbs showing impaired flexion at the interphalangeal joints.

The mother, who was 147 cm tall, recalled being told that her own anterior fontanelle had not closed until the age of 5 years. On examination this lady, who was of limited intellect and experienced difficulty in reading and writing, had a flat receding forehead, bilateral proptosis, right ptosis, prominent crura of both ears, short fingers, and broad halluces. Palpation of her skull revealed bilateral parasagittal parietal defects each admitting a finger tip. Skull x-ray (fig 2) confirmed the presence of bilateral parietal foramina with prominent digital markings and a large pituitary fossa.

The findings in these patients and those of Dr Thompson and colleagues illustrate the curious and important point that a disorder traditionally associated with craniosynostosis may present paradoxically with delay in closure of the fontanelles and sutures. The fact that our patients showed limited intellectual development suggests that retardation in the Saethre-Chotzen syndrome may be a primary effect of the gene rather than a result of craniosynostosis with raised intracranial pressure, as has been suggested.2

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References

Philtrum length, intercommissural distance, and ear measurements in newborn infants

SIR,

We thank Dr Mehes for his important comment about our measurements in newborn infants.1 We agree indeed that all anthropometric measurements should be evaluated in relation to birth weight also. This is especially important when birth weight is not appropriate for gestational age, as in such cases.
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some of the body measurements and proportions might deviate from normal. Also, for the immature preterm infant, the use of normal values in relation to weight allows anthropometric evaluation in the neonatal period beyond the first week of life (birth to 41 weeks conceptual age).

In order to allow the clinician to use anthropometric measurements of philtrum length and intercommissural distance and to define normal ear length and low set ears in these infants, we present normal values for these parameters in relation to birth weight (figs 1 to 5). The measurements were obtained from the data previously described in this journal1–3 using the same study population, methods, definitions, and analysis. The infants were divided into birth weight groups (751 to 1000 g; 1001 to 1250 g; 1251 to 1500 g, etc) and the mean ± 2 SD for each group was used.

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References