

## Answers to the Medical Quiz

- A1. The skin spots shown in Fig 1 and Fig 2 are cafe au lait patches.
- A2. About 10% of normal people have 1 to 5 cafe au lait spots of more than 1.5 cm in diameter.
- A3. Besides a positive history, diagnosis of neurofibromatosis (Von Recklinghausen's disease) rests upon presence of these skin patches in 75% of the cases. Incidentally, family history for neurofibromatosis was positive in our patient.

Diagnosis of neurofibromatosis (Von Recklinghausen's disease) rests upon a positive family history and the skin manifestations, usually consisting of more than 6 cafe au lait spots larger than 1.5 cm in diameter. As early as 1964 Crowe found that 10% of 6850 normal people had 1 to 5 cafe au lait spots of more than 1.5 cm<sup>1</sup>. In 1966, Whitehouse studied children less than 5 years of age and found only 0.75% of normal children had more than 2 spots. Five spots or more measuring at least 0.5 cm

each make the diagnosis mandatory<sup>2</sup>, in the paediatric age group. Axillary freckling associated with cafe au lait patches is also a strong presumptive evidence of neurofibromatosis<sup>3</sup>.

The cafe au lait patches are flat lightly pigmented, non-hairy spots; are multiple over the trunk and limb in neurofibromatosis. They increase in number and size as the child gets older but behave usually more like hamartomas and do not become malignant. In contrast, fibromas of neurofibromatosis are small pink pedunculated skin tumours which usually appear only at puberty.

### REFERENCES

1. Crowe FW. Axillary freckling as a diagnostic aid in neurofibromatosis. *Ann Intern Med* 1964;61:1142.
2. Whitehouse D. Diagnostic value of the cafe au lait spot in children. *Arch Dis Child* 1966;41:316.
3. Brown JK. Leucodystrophies and leucoencephalopathies (Disorders of myelination). In: Forfar JQ, Arneil GC, eds. *Textbook of Paediatrics*. 2nd ed. Edinburgh: Churchill Livingstone, 1978:691-2.