

ANSWER TO PHOTO QUIZ (PAGE 43)

PANCYTOPENIA IN A YOUNG GIRL WITH SKIN LESIONS

DIAGNOSIS

The haematological condition is acute myeloid leukaemia and the syndrome in this patient is tuberous sclerosis. Tuberous sclerosis is an inherited neurocutaneous syndrome characterised by multiple benign hamartomas involving brain, eyes, heart, lung, liver, kidney, and skin.¹ It has an autosomal dominant pattern of inheritance with an approximate incidence of 1 in 5000 to 10,000 live births.² Tuberous sclerosis can be diagnosed clinically using the criteria from the International Tuberous Sclerosis Complex Consensus Conference.³ Patients with tuberous sclerosis are at an increased risk of developing malignant tumours involving kidneys, brain, and soft tissues.⁴ The risk of malignancy in tuberous sclerosis is approximately 18-fold higher than in the normal population. Downregulation of tuberous sclerosis complex 2 expression has been demonstrated in acute myeloid leukaemia.⁵

REFERENCES

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