Severe short stature with features of achondroplasia, later diagnosed as panhypopituitarism - a case report

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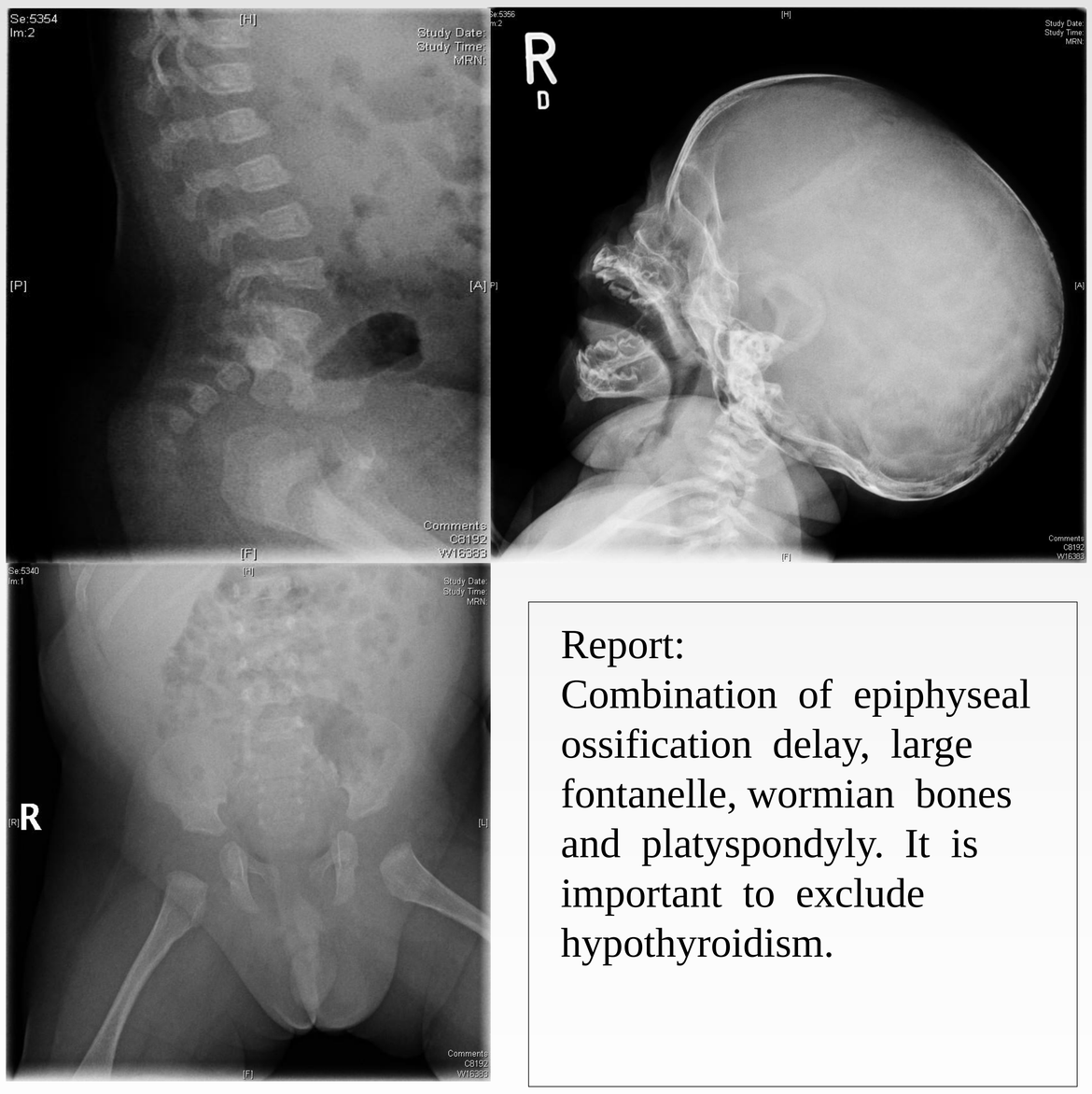
BACKGROUND

Neonatal screening for congenital hypothyroidism in the UK will not detect central hypothyroidism with low TSH. However, late diagnosis is uncommon as children with hypopituitarism are likely to present with other pituitary abnormalities. Skeletal dysplasia can be associated with severe hypothyroidism[1].

CASE PRESENTATION

- Male infant born at 31 weeks gestation with birth weigh of 1.935 kg
- No documented hypoglycaemia in postnatal period
- Subsequent failure to thrive with short stature
- At 8 months, his weight was 5.2 kg (-5 SDS), length 55.5 cm (-5 SDS) with head circumference 44.5 cm (50th centile)
- He appeared to have a disproportionate skeletal appearance with proximal shortening leading to request for a skeletal survey
- No other obvious dysmorphic features
- Normal phallus; undescended right testes

SKELETAL SURVEY



Report:
Combination of epiphyseal ossification delay, large fontanelle, wormian bones and platyspondyly. It is important to exclude hypothyroidism.

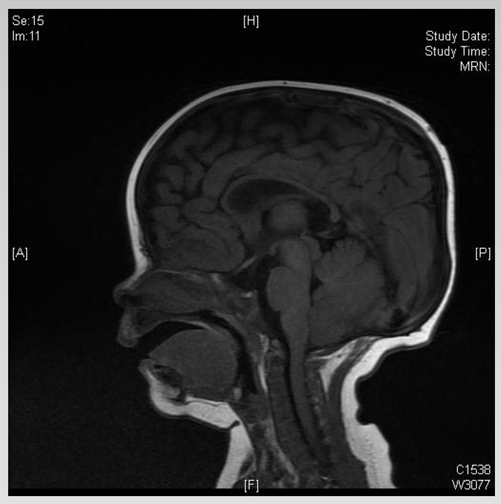
FURTHER INVESTIGATIONS

Age 10 months
- TSH 1.13 IU/l; free T4 of 3.6 pmol/l – commenced on levothyroxine 25mcg
- Synacthen: Peak cortisol 345nmol at 30 mins – commenced on hydrocortisone 1mg tds
- ACTH 8.82ng/L (10 – 50)
- IGF 1 <25ng/ml (51-303); IGFBP3 <0.5mg/l (0.8-3.9)

- LHRH test

Time	0h00m	0h20m	1hr00m
LH (IU/L)	<0.2	2.9	1.8
FSH (IU/L)	<0.2	0.2	0.3
Testosterone (nmol/L)	<0.69		

- MRI brain:
  small anterior pituitary
  normal post pituitary



- Subsequent collapse with hypoglycaemia. Documented repeated hypoglycaemia which resolved on commencing growth hormone

CONCLUSIONS

- No screening programme is perfect and as in this case, false-negative screening results can occur. [1]
- Thyroid function should be checked in all cases of short stature and suspected skeletal dysplasia.
- Funding for GH treatment can be difficult to secure. In this situation, hypoglycaemia was managed with GH treatment and funding secured on emergency basis.

REFERENCES

1. Hüffmeier U, Tietze, HU, Rauch, A: Severe skeletal dysplasia caused by undiagnosed hypothyroidism, Eur J Med Genet 2007 May-June ; 50(3): 209-15