Familial Glucocorticoid Deficiency

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Background

Familial glucocorticoid deficiency (FGD) is a rare autosomal recessive disorder associated with isolated glucocorticoid deficiency. Melanocortin−2 receptor (MC2R), account for approximately 25% of FGD cases

Case Study

A 3 year old girl presented with recurrent hypoglycaemic episodes from day 2 of life. She was a product of consanguineous family born with a birth weight of 2.3 kg. She was found to be dark in complexion from birth. Hyoglycaemic convulsions were noted on day 2 of life. Thereafter she had 3 more episodes of convulsions associated with hypoglycaemia. All 3 episodes were associated with respiratory tract infections associated with wheezing where she required hydrocortisone treatment. She had delay in achieving gross motor milestones. She was at +1SD for her height and -1SD for her weight (Figure 1.)

Test	Result
DHEAS	<0.078 µmol/l
Aldosterone	15.74pg/ml (12-340)
Renin	40 pg/ml
Testosterone	0.662pmol/l
fT4	12.71pmol/l
TSH	2.2mIU/l
17OHP	<0.15nmol/l
ACTH	6320 pg/ml

Table 1.

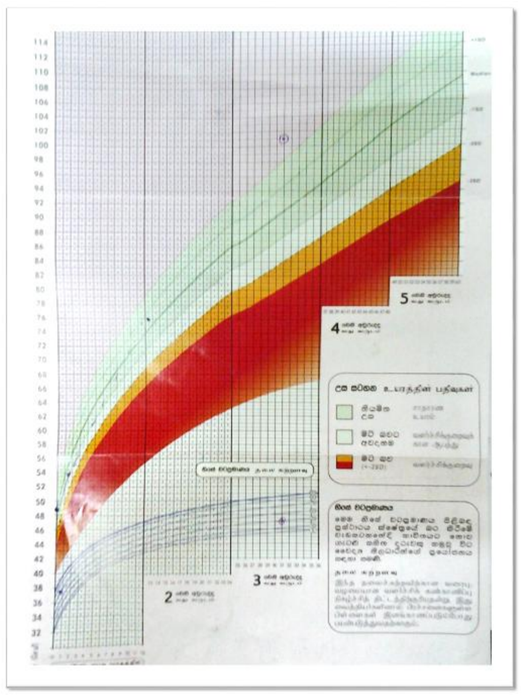

Figure 1.

Synacthen Test	Cortisol
0 min	<41nmol/l
30 min	<41nmol/l
60 min	Sample Insufficient

Table 2.

Her genitalia were of normal female.

Karyotype was 46, XX.

Bone age was 7 years at the chronological age of 3 years.

Results of the biochemical tests are given in the Table 1 and Table 2.

There were no clinical features to suggest Allgrove syndrome.

Hydrocortisone replacement therapy was started after investigations. Pigmentation reduced with treatment (Figure 2.)

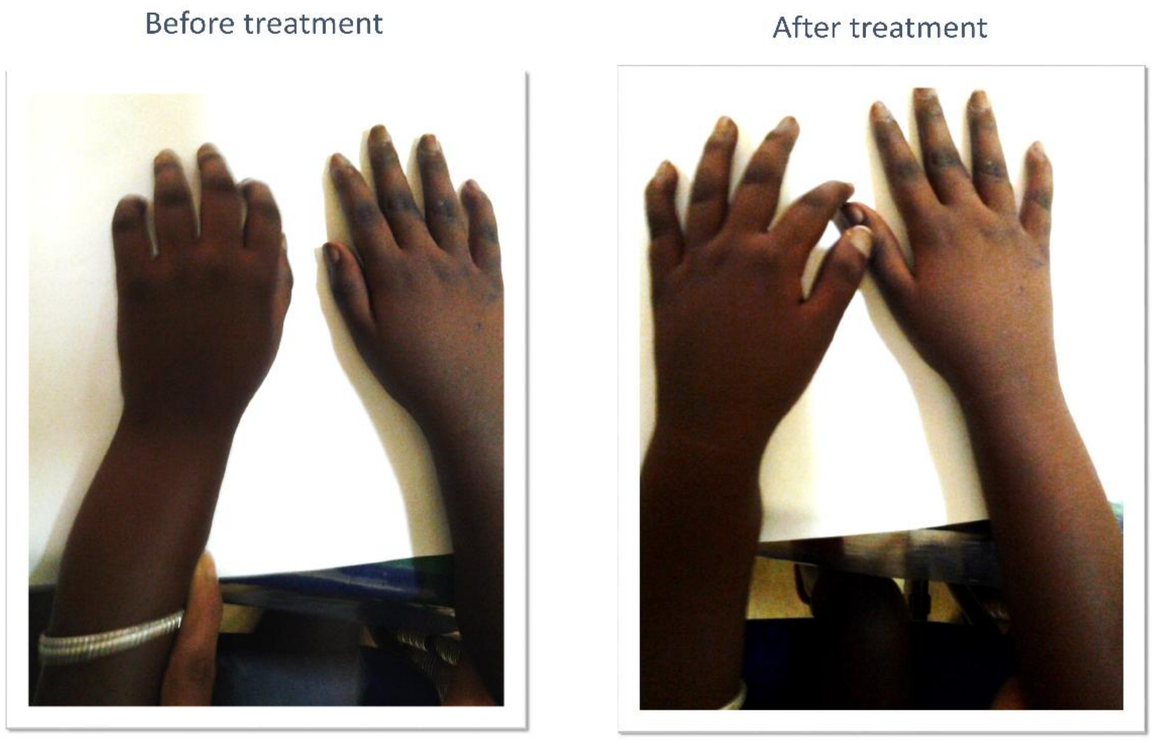

Figure 2

Conclusion

Possibility of MC2R mutation was considered due to advanced bone age together with glucocorticoid deficiency. Her diagnosis could have been considered at birth itself when she presented with hypoglycaemia together with dark complexion. Hydrocortisone treatment given during respiratory tract infections with wheezing would have saved the child with acute episodes. Cortisol deficiency has to be excluded when children present with recurrent hypoglycaemia.

References

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