

Hypoglycemia in a patient with Turner syndrome and Kabuki make-up

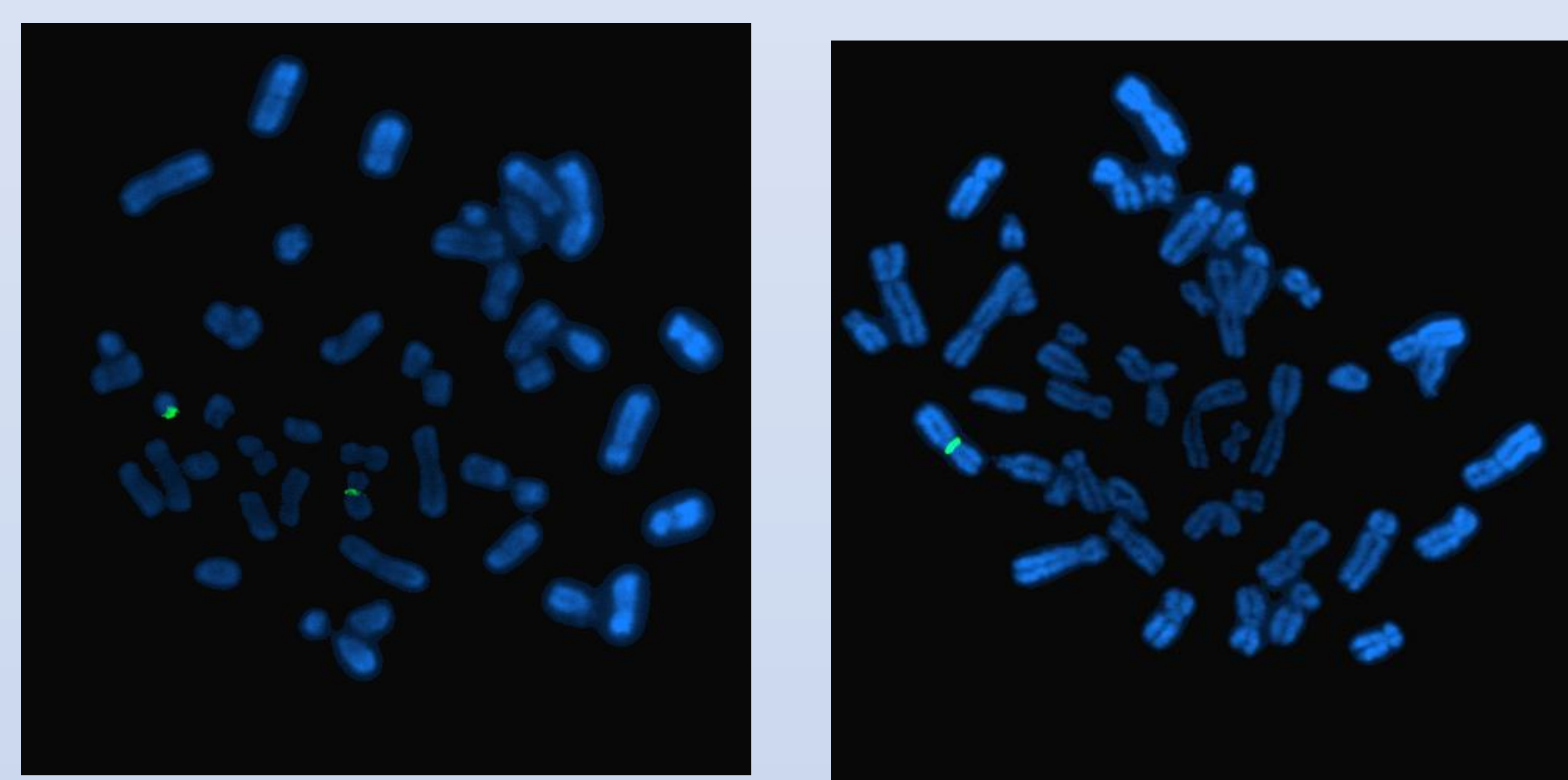
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Introduction: We report on the case of a 1 year old girl with a complex mosaic Turner syndrome and Kabuki-like phenotype. She presented mild hyperinsulinemic hypoglycemia. Despite the rareness of this case, clinicians should be aware of the possibility of hyperinsulinemic hypoglycemia in patients with Turner syndrome

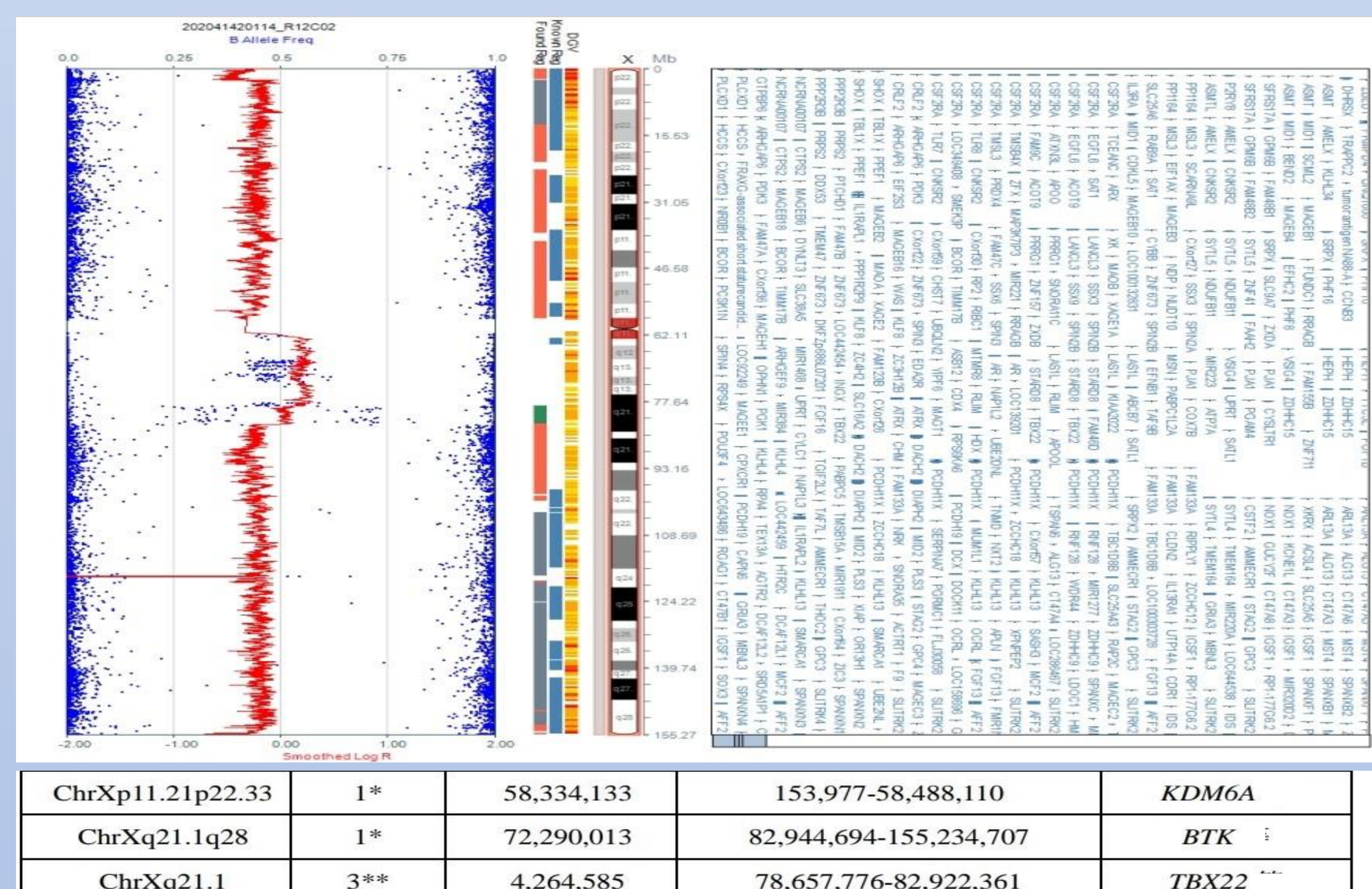
Case presentation

- 1 years old girl first admitted to our hospital because of seizure afebrile at 6 months old and was found extremely hypoglycemia with a blood glucose concentration of 1.8mmol/l
- Birth weight was 2900g (10th–25th centile), birth length was 48cm (10th– 25thcentile).
- Both parents are in good health. The family history was unremarkable.
- She She had special facial features reminiscent of the Kabuki syndrome

FISH:mos 46,X,+mar [50] /45,X[50].ish mar(X)(DXZ1+)



DXZ1 (Xp11.1-q11.1, green) SRY (Yp11.31-p11.32, yellow)

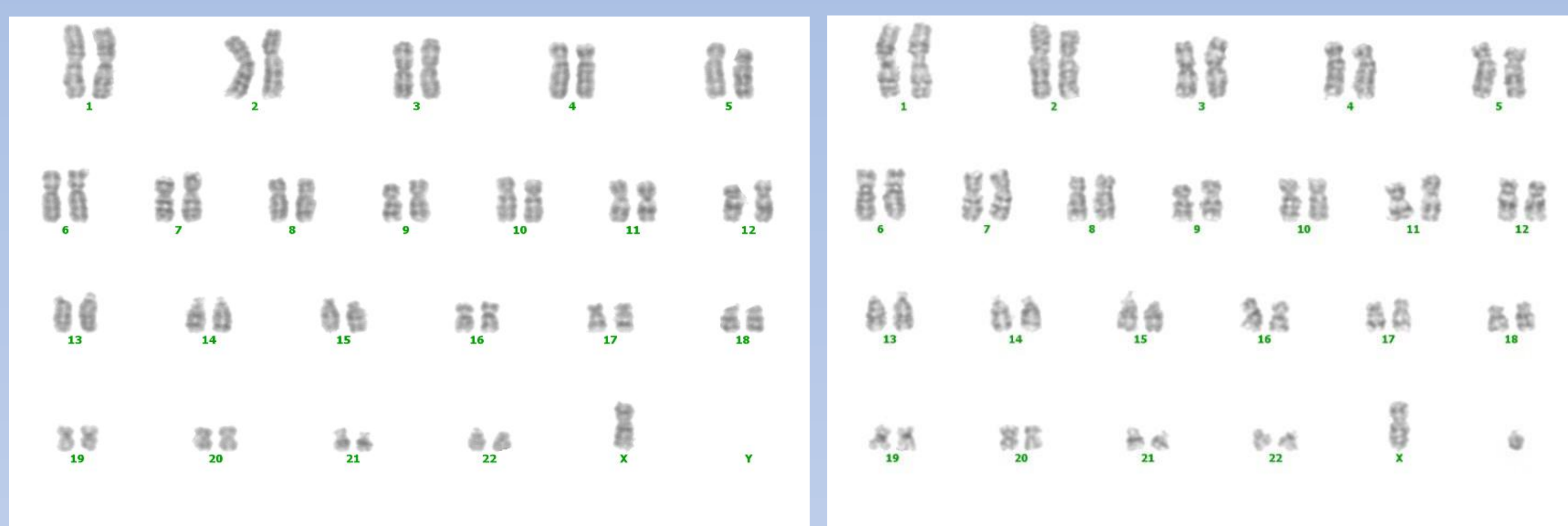


Investigation

- Results of the hormones at the time of hypoglycemia (fasting test)

Hormones	Value	Reference
Plasma glucose(mmol/L)	1.8	3.5-5.5
Serum insulin(mU/L)	2.9	<2
Serum cortisol(mmol/L)	381	<500
adrenocorticotrophic hormone (pg/mL)	202	---
Chromosome karyotype(45,X/mos46,X,+mar; Serum growth hormone (ng/ml)	5.51	---
serum free thyroxine(ug/dl)	12	10.8-20
HbA1C(%)	4.2	4-6

Chromosome karyotype: 46,X,+mar /45,X.



References

- Alkhayat H, Christesen HB, Steer J, Stewart H, Brusgaard K, Hussain K: Mosaic Turner syndrome and hyperinsulinaemic hypoglycaemia. *J Pediatr Endocrinol Metab* 2006;19:1451–1457.
 Stankiewicz P, Thiele H, Giannakudis I, Schlicker M, Baldermann C, Kruger A, et al. Kabuki syndrome–like features associated with a small ring chromosome X and XIST gene expression. *Am J Med Genet.*2001;102:286–92.

CONCLUSION

- hypoglycemia should be suspected in patients in mosaic Turner syndrome with presenting with seizures and other nonspecific symptoms.
- The mechanism leading to HH and Kabuki make up in this condition is still unknown.
- *KDM6A* may play a role in it.