



An infant with X-linked adrenal hypoplasia congenita and Xp21 contiguous gene deletion syndrome

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Background

Contiguous gene syndromes are disorders caused by deletions of genes that are adjacent to one another. It is caused by partial deletion of Xp21, which includes the genes responsible for glycerol kinase deficiency, congenital adrenal hypoplasia, Duchenne muscular dystrophy (DMD) and intellectual disability. We report the case of a 14-day-old patient with this rare disease.

Case Report

A 14-day-old newborn was referred to our clinic for scrotal hyperpigmentation. He was born at 40 weeks of an uneventful pregnancy. His parents were first-degree cousins. Family history was unremarkable. On examination; blood pressure was 70/39 mmHg and his testes were 2 mL in volume bilaterally and he had scrotal hyperpigmentation, hyponatremia, hyperpotasemia with elevated ACTH (485 pg/mL) and cortisol 8.48 µg/dL suggesting adrenal insufficiency. Karyotype was normal. Hydrocortisone and flornidol treatments were started. When he became 2 months of age, he was hypotonic and he had high creatine phosphokinase (2111 IU/L) and high triglyceride (732 mg/dL) levels suggesting DMD and dystrophin gene deletion was detected. We suspected contiguous gene deletion syndrome in Xp21 and SNP microarray analysis was performed using CytoSNP12 microarrays and detected a 3,982,668 bp deletion of the X chromosome, ranging from p21.2 to p21.1 regions with minimal extent nt: 29,555,683-33,538,351 (Figure 1) comprising the genes responsible for DMD, glycerol kinase deficiency, mental retardation (IL1RAPL1), and the congenital adrenal hypoplasia (gene DAX1 or NROB1 gene: Xp21.2-21.1).

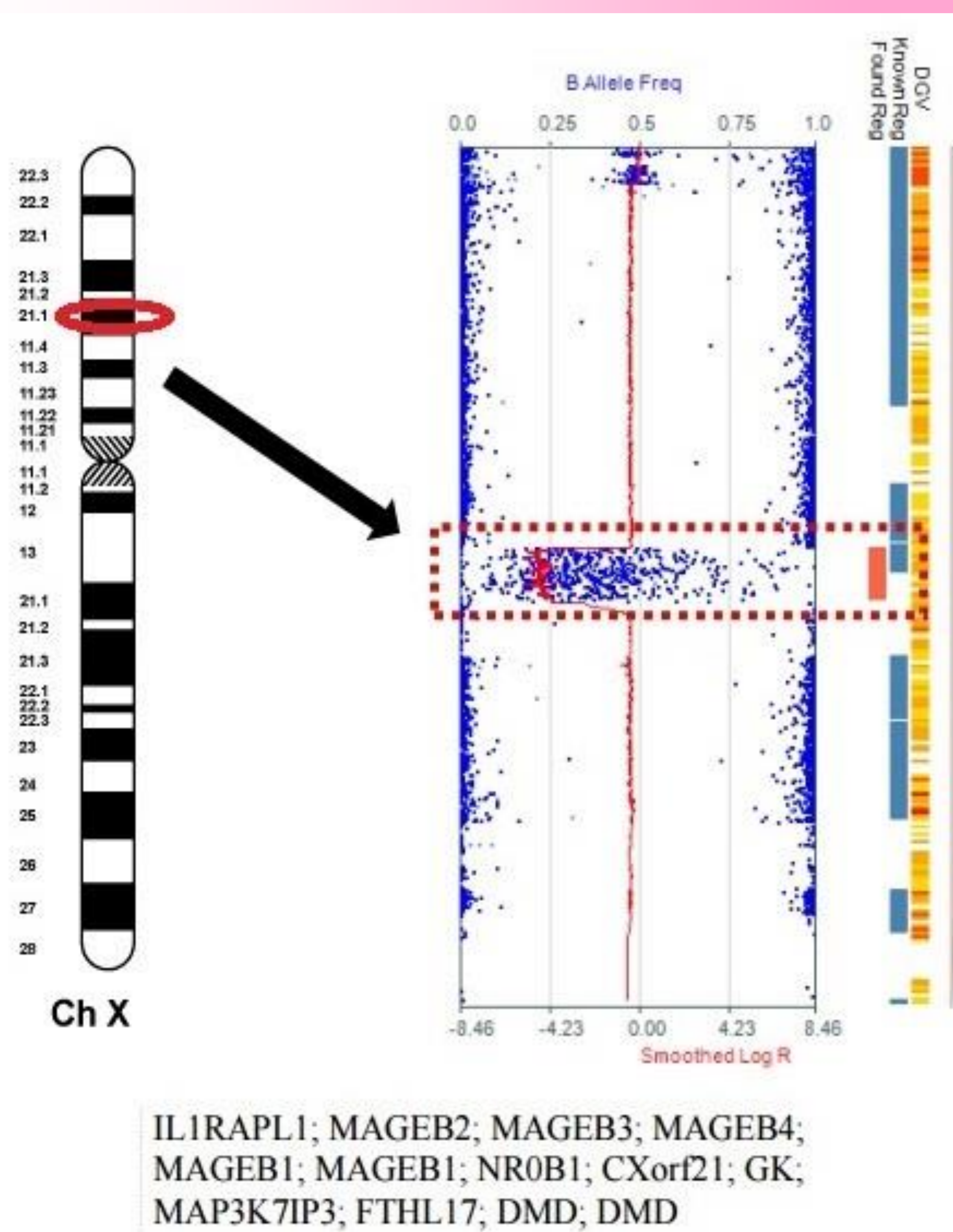


Figure 1: Deletions of Xp21 (from p21.2 to p21.1) and comprising genes (CytoSNP12 microarrays; Illumina, San Diego, CA, USA).

References

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Conclusion

The Xp21 contiguous gene deletion syndrome should be considered in any infant with adrenal insufficiency. Symptoms depend on the size of deletion and appear almost exclusively in the male gender. Usually the first and most severe are the signs of adrenal hypoplasia. Measurement of serum triglycerides and creatine kinase activity are simple screening tests that may facilitate early diagnosis and appropriate genetic counseling about risks of recurrence in subsequent offspring.

Nothing Disclosure

