Recurrent hypoglycemia due to growth hormone deficiency in an infant with Turner syndrome.

Growth hormone (GH) deficiency may occur in Turner syndrome (TS), but infantile hypoglycemia attributable to TS with GH deficiency has not been reported before. We report a puzzling case of neonatal hypoglycemia due to GH deficiency in Turner syndrome. Array CGH was used to scrutinize the complex TS karyotype. Standardized laboratory procedures. In a preterm (32 weeks) with prolonged and cholestatic jaundice, recurrent hypoglycemia occurred at the age of 1.5 months and was related to GH deficiency. There were no other endocrine or syndromic features. GH therapy was started at a usual dose of 25-30 µg/kg/day, but hypoglycemia recurred. Hepatopathy and hypogammaglobulinemia suggested X-recessive GH deficiency type 3 with non-random X-inactivation but resolved spontaneously. Nonetheless, a 45,X[75]/46,X,i(Xq)[21]/47,X,i(Xq)x2[4] TS karyotype was diagnosed with an apparent isochromosome fusion at the centromere. Upon this diagnosis, GH dose was doubled (50 µg/kg/day), and blood glucose was normalized consistently. In array CGH, the signal of Xp deviated more strongly than that of Xq, but the relation of the signals differed substantially from what the karyotype predicted. The isochromosome fusion point was relocated to Xp11.22, distal to a block of mental retardation genes that escape X-inactivation. TS with GH deficiency should be considered as a
potential differential diagnosis of hypoglycemia in infants requiring higher doses of GH. ii) While array CGH may be erroneous in quantification of TS mosaicism, it is useful in precisely delineating isochromosomes and identifying genes on them that escape X-inactivation and thus possibly affect the TS phenotype.