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Early diagnosis of Duchenne muscular dystrophy in 6-months-old male with primary adrenal insufficiency

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BACKGROUND

Adrenal hypoplasia congenital associated with *DAX-1* (*NROB1*) gene mutations is a rare cause for primary adrenal insufficiency in male. It can be presented as a part of Xp21 contiguous gene deletion syndrome which characterized by complex glycerol kinase deficiency (*GK*), adrenal hypoplasia congenital (*NROB1*), intellectual disability (I*L1RAPL1*) and/or Duchenne muscular dystrophy (*DMD*)

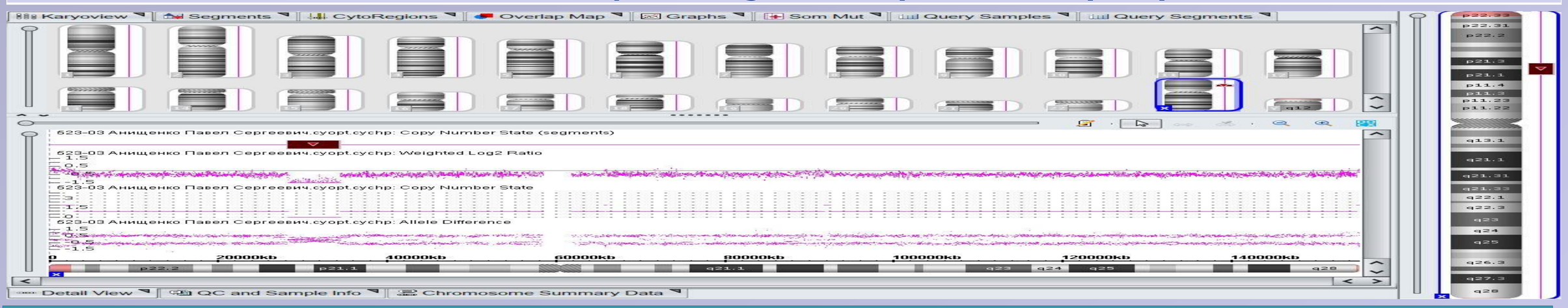
CASE REPORT

We report a 6-month-old male infant with primary adrenal insufficiency, unilateral cryptorchidism and high levels of transaminases. An adrenal insufficiency was diagnosed at 6 weeks after birth due to low weight gain, hyperpigmentation, hypotonia, hyponatremia, hyperkalemia, high levels of ACTH and renin. Evaluation at 6 months of age revealed elevated liver enzymes, increased levels triglyceride and extremely increased creatine phosphokinase (Tab.1). It was suggestive for myodystrophy. We supposed that the combination of adrenal insufficiency with cryptorchidism, and laboratory signs of myodystrophy could result from the Xp21 contiguous gene deletion syndrome. Further investigations showed massive glyceroluria, and the glycerol kinase deficiency was suspected. Microarray analysis showed the Xp microdeletion in Xp21.2-p21.3(28332614_34432348) loci, which involved *IL1RAPL1, NR0B1, GK, DMD* genes (Pic.1). During last examination at the age of 6 month the mental retardation and/or symptoms of muscular dystrophy were not seen.

Table 1. Laboratory data of male patient with Xp21 contiguous gene deletion syndrome

NR0B1 –X-linked congenital adrenal hypoplasia with hypogonadotropic hypogonadism	***GK*** **-** Glycerol kinase deficiency	***DMD*** **-** Duchenne muscular dystrophy
Before treatment: Na 125 mmol/l (135-145) K 8 mmol/l (3.5-5.1)	Triglyceride 5,18 mmol/l (0,1-1,7)	ALT 281 U/L (0-55), AST 275 U/L (5-34)
ACTH 293 pg/ml (7-66)	Glycerol (urine) 482,2 mM/M (0-9)	Creatine phosphokinase 15000 U/L (30-200)
Renin >500 U/l (2.8-39.9)		

Picture 1. Comparative genomic hybridization (CGH)



CONCLUSIONS

Congenital adrenal hypoplasia is a rare disease that could be associated with of Xp21 contiguous gene deletion syndrome. Boys with this disorder should be screened for other components such as complex glycerol kinase deficiency, Duchenne`s muscular dystrophy and mental retardation. Early diagnosis of Duchenne`s muscular dystrophy can be helpful for appropriate treatment strategy and prenatal diagnosis in the family.