

SIX CASES OF CONGENITAL HYPERINSULINISM CAUSED BY **MUTATIONS IN MODY GENES**



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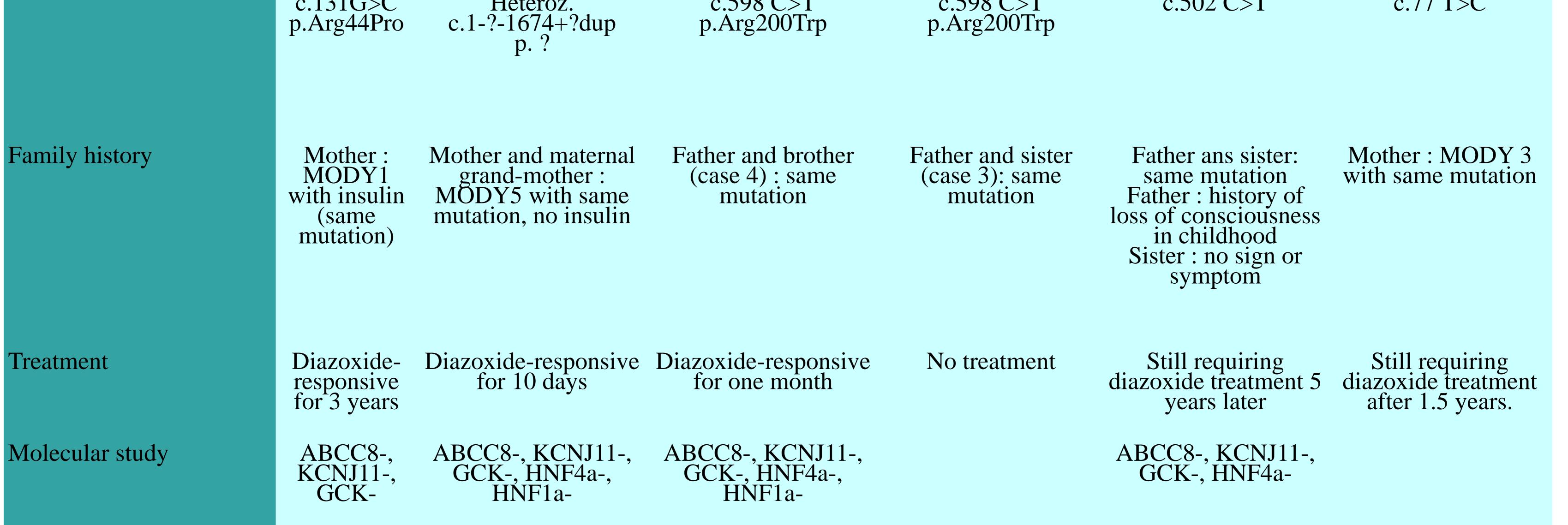
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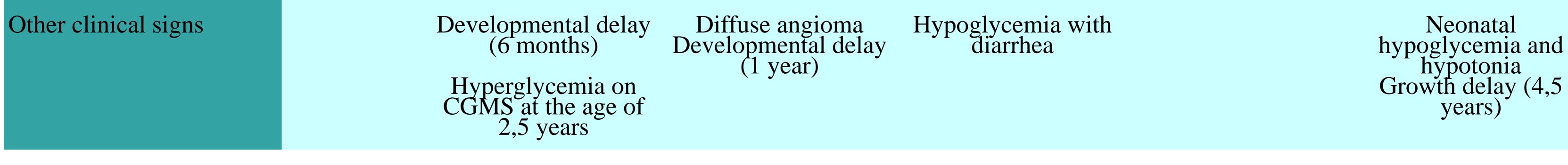
INTRODUCTION

Congenital hyperinsulinism (HI) is the most common cause of persistent hypoglycemia in infants and children (1). Recently, mutations in genes usually involved in MODY 1 and 3 have been described in HI (2). We present here 6 cases of hyperinsulinism associated with MODY1 (1 case) and MODY3 (4 cases) and 1 case of HI associated with MODY5 mutation.

CASES REPORT

	CASE 1	CASE 2	CASE 3	CASE 4	CASE 5	CASE 6
Sex (year)	GIRL (2011)	BOY (2013)	GIRL (2012)	BOY (2015)	BOY (2004)	GIRL
Gestational age (wks)	35+5	41	38+1	39+2	34+3	39
Birth weight (g) Birth length (cm) Head circumference (cm)	3350 47,5 33	3670	2610 41 34	2990 50 35,5	1570 39	normal
Diagnosis of hypoglycemia and HI due to MODY mutation	Neonatal	Neonatal Transient hyperinsulinism	Neonatal	Neonatal	4,7 years Hypoglycemia seizure with ketonuria	5 years Hypoglycemia seizure with ketonuria
Mutation	exon 2 of HNF4a gene Heteroz. c.131G>C	exon 1-9 of HNF1b gene Wide duplication Heteroz.	Exon 3 of HNF1a gene Heteroz. c.598 C>T	Exon 3 of HNF1a gene Heteroz. c.598 C>T	HNF1a gene	HNF1a gene





CONCLUSION

We report the first case of HI associated with HNF1b mutation. HNF4a and HNF1a are recently described causes of HI. Our cases showed that the clinical presentation can be variable, from transient neonatal HI to persistent HI discovered during childhood.



1. Arnoux JB, Congenital hyperinsulinism: current trends in diagnosis and therapy. Orphanet J Rare Dis. 2011

Neonatal

years)

2. Stanescu DE, Novel presentations of congenital hyperinsulinism due to mutations in the MODY genes: HNF1A and HNF4A. J Clin Endocrinol Metab 2012

