



A hyperinzulinizmosos hypoglycaemia „a fordított diabetes”

Dr Luczay Andrea

J Pediatr Surg. 2002 Feb;37(2):155-8.

Partial elective pancreatectomy is curative in focal form of permanent hyperinsulinemic hypoglycaemia in infancy: A report of 45 cases from 1983 to 2000.

Crétolle C¹, Fékété CN, Jan D, Nassogne MC, Saudubray JM, Brunelle F, Rahier J.



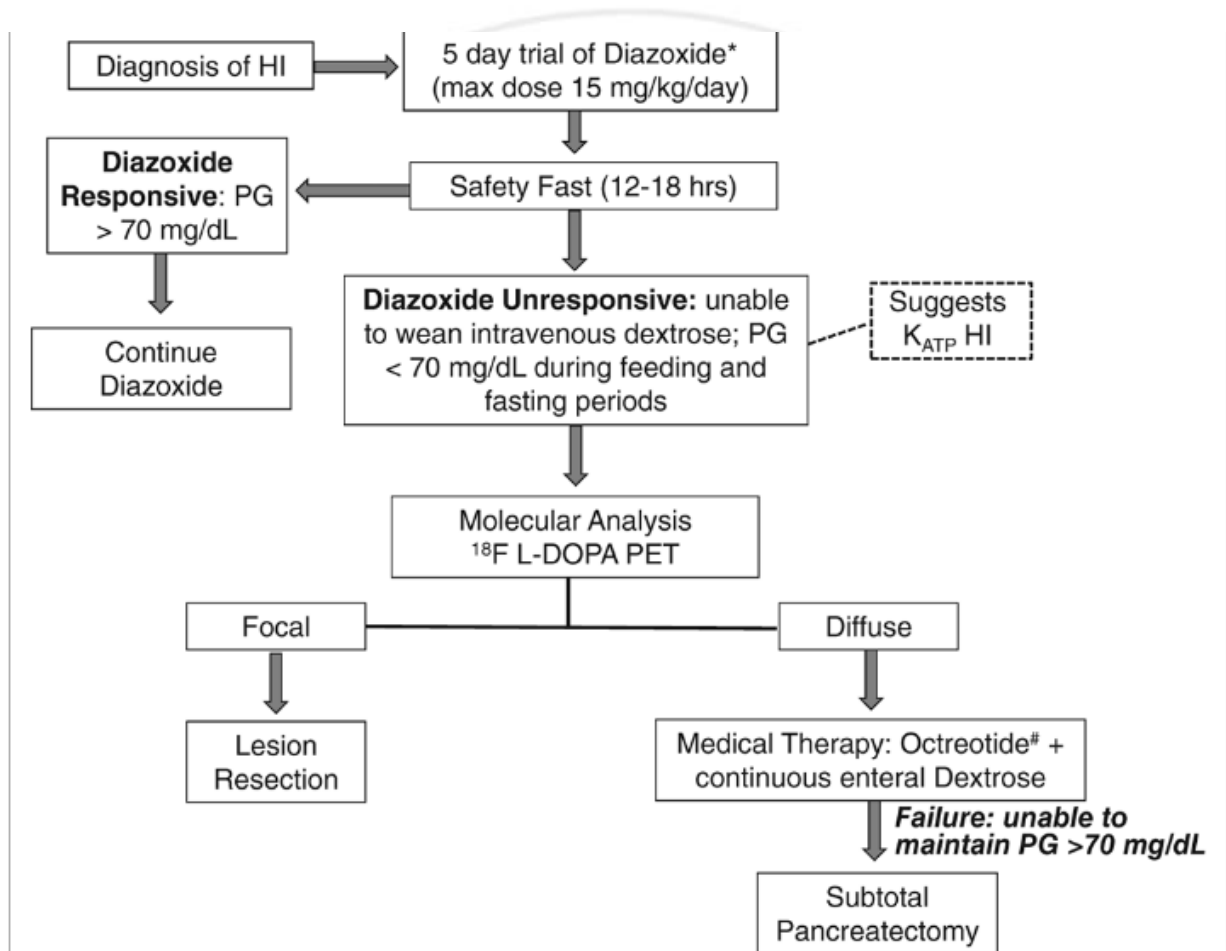
Severe Hyperinsulinemic Hypoglycemia
in a Neonate: Response to Sirolimus
Therapy.

Méder Ü, Bokodi G, Balogh L, Körner A,
Szabó M, Pruhova S, Szabó AJ.

Pediatrics. 2015 Nov;136(5):e1369-72.
doi: 10.1542/peds.2014-4200.

	Gene	Locus	Mode	Triggers	Diazoxide Response	↑NH ₃
Monogenic HI						
KATP-Channel (SUR1/Kir6.2 subunits)	ABCC8/KCNJ11	11p15.1				
Recessive KATP-HI	ABCC8/KCNJ11	11p15.1	Rec	F/P	-	
Dominant KATP-HI						
Diazoxide-unresponsive	ABCC8 (only?)	11p15.1	Dom	F/P	-	
Diazoxide-responsive	ABCC8/KCNJ11	11p15.1	Dom	F/P	+	
Focal KATP-HI (pat. 11p UPD + pat. rec. KATP mutation)	ABCC8/KCNJ11	11p15.1	Sporadic	F/P	-	
GDH	GLUD1	10q23.3	Dom	F/P/Leu	+	+
GCK	GCK	7p13	Dom	F	-	
SCHAD	HADH1	4q25	Rec	F/P/Leu	+	
UCP2	UCP2	11q13.4	Dom	F/Glu	+	
HNF4A	HNF4A	20q13.12	Dom	F	+	
HNF1A	HNF1A	12q24.31	Dom	F	+	
Pyruvate transporter (MCT1)	SLC16A1	1p13.2	Dom	Exercise	-	
HK1	HK1	10q22.1	Dom	F/Glu	+	
PGM1	PGM1	1p31.3	Rec	Glu	-	

Stanley CA J Clin Endocrinol Metab. 2016 Mar; 101(3):815-826



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Congenital Hyperinsulinism Center

Pediatr Diabetes 2017 Feb;18(1):3-9.

Terápiás lépések

Diazoxid 20 mg/kg/nap po
(3 részre)

Hypothiazid 0,7-1 mg/nap

Sandostatin 35 ug/kg/ nap sc
(4 részre)

Sandostatin LAR im
31 napi adag 4 hetente

Sirolimus
0,5 mg/m²/nap p.o

Szérumszint:
5-15 ng/ml
(25-30 ng/ml)

CÉL: $vc > 3,6$ mmol/l min 6 óra éhezés után is

	Feeding problems (<i>n</i> = 28)	No feeding problems (<i>n</i> = 55)	<i>p</i> value
Age at presentation (days)	1 (1–365)	1 (1–630)	0.21
Insulin at diagnosis (mU/l)	18.5 (2.5–110.0)	11.2 (2.2–132.0)	0.32
CHI mutations (%)	17 (63.0)	15 (26.8)	0.002 [*]
Maximal dose of diazoxide (mg/kg/day)	15.0 (7.0–20.0)	6.1 (4.5–21.0)	<0.001 [*]
Octreotide treatment (%)	6 (22)	0 (0)	<0.001 [*]
Glucagon infusion (%)	14 (51.9)	5 (8.9)	<0.001 [*]
Subtotal pancreatectomy (%)	10 (35)	0 (0)	<0.001 [*]
Focal CHI (%)	5 (18.5)	3 (5.4)	0.06
Spontaneous resolution (%)	6 (22.2)	32 (58.2)	0.002 [*]

Front Endocrinol (Lausanne)2016; 7: 8



Saját betegeink

- 11 beteg 4 lány életkor: 6 hét 13 év
- 2 fiú hyperammonaemiás hypoglycaemia

Diazoxid +

- 7/9 genetikai vizsgálat
 - 1 ABCC8 compound het
 - 2 ABCC8 apai eredetű heterozig
 - 1 KCJN11
 - 2 ABCC8 heterozigota de novo
 - 1 nem talált mutációt

Saját betegeink 2

- Diazoxid + 3/9
- Sandostatin LAR 3/9
- Sirolimus 1/9

PET: 5 esetben 2 focalis

1 sikeres műtét Dániában

1 sikeres műtét Budapesten

PEG:5 esetben

Kimenetel

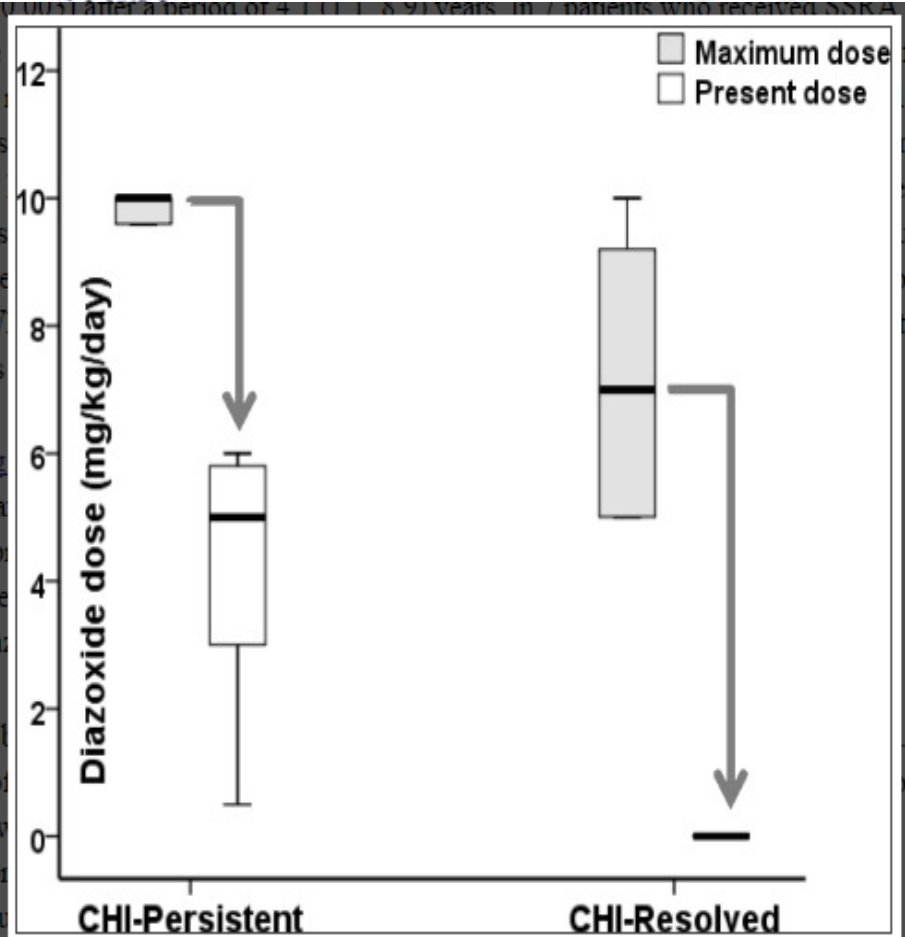


Fig. 1
 Maximum and present doses of diazoxide in children with CHI represented as box and whisker plots (median, 95% confidence intervals). In persistent CHI (CHI-Persistent), a higher maximal dose of diazoxide was required than in patients with resolved CHI ...

Teljes gyógyulás

8/14 Diazoxid 1.6 év

7/7 Sa 5.8 év

(3,1 év)

8/21 neurológiai érintettség

