

Congenital adrenal hyperplasia in adults

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Congenital adrenal hyperplasia (CAH) is the commonest genetically determined endocrine disorder with more than 95 % cases caused by the mutation in gene for 21-hydroxylase. We have consensual guidelines for the care about children with CAH but not for adults. Till now, there are only few studies mapping health status in sufficiently large groups of adult CAH patients. The aim of our work was to examine health status in Czech CAH patients from several centres which cover of about 40-50 % of the Czech Republic.



Methods

Lipids, blood glucose, blood pressure and anthropometrical data in 31 males and 71 females with CAH (classic form, deficiency of 21-hydroxylase) were compared with data from the study Czech post-MONICA (1% random population sample). CAH patients were examined during their routine exam at different centers with the exc. of DXA. The results are stated as age-adjusted mean and SEM, p value is from t-test.

Metabolic parameters

	Men		p
	CAH (N=31)	Post MONIKA (N=1361)	
age (yrs)	29,3 (11,8)	44,7 (10,2)	
height (cm)	163,8 (1,3)	178,5 (0,19)	0,001
weight(kg)	72,3 (2,93)	89,3 (0,43)	0,001
BMI (kg/m ²)	27,09 (0,84)	28,01 (0,12)	
systolic TK (mmHg)	128,75 (3,01)	130,9(0,43)	
diastolic TK (mmHg)	82,4 (1,8)	84,4 (0,27)	
cholesterol (mmol/l)	4,62 (0,23)	5,21 (0,03)	0,009
HDL-cholesterol (mmol/l)	1,51 (0,08)	1,30 (0,01)	0,004
LDL-cholesterol (mmol/l)	2,78 (0,21)	3,19 (0,03)	
triglycerides (mmol/l)	1,16 (0,18)	1,61 (0,02)	0,018
blood glucose (mmol/l)	5,56 (0,23)	5,50 (0,03)	
osteopenie/porosa	5/14 (35 %)		
cholesterol >5	24%		
LDL-cholesterol >3	24%		
obesity (BMI >30kg/m ²)	26%		

	Women		p
	CAH (N=71)	Post MONIKA (N=1561)	
age (yrs)	32,4 (10,8)	43,9 (9,9)	
height (cm)	156,6 (0,8)	165,7 (0,2)	0,0001
weight(kg)	69,2(1,95)	72,8 (0,39)	
BMI (kg/m ²)	28,5 (0,7)	26,5(0,14)	0,005
systolic TK (mmHg)	128,1(2,02)	124,4 (0,4)	
diastolic TK (mmHg)	79,5 (1,16)	80,14 (0,23)	
cholesterol (mmol/l)	5,48 (0,13)	5,16 (0,02)	0,016
HDL-cholesterol (mmol/l)	1,81 (0,06)	1,63 (0,01)	0,002
LDL-cholesterol (mmol/l)	3,07 (0,14)	2,98 (0,02)	
triglycerides (mmol/l)	1,23 (0,09)	1,23 (0,02)	
blood glucose (mmol/l)	5,05 (0,13)	5,15 (0,02)	
osteopenie/porosa	8/26 (31 %)		
cholesterol >5	56%		
LDL-cholesterol >3	41%		
obesity (BMI >30kg/m ²)	23%		

Parameters of compensation and mode of treatment

17 OHP > 30 nmol /l 41,2 % patients

< 10 nmol/l 28,4 % patients

Hydrocortison 52 %, dexametason 25 %, prednison 18 % , prednison + hydrocortison 3 %

Discussion and Conclusions

When we supposed CAH frequency as 1: 15000, the cohort under study comprised only 15 % of supposed CAH patients. It means that the majority of patients is not treated in the specialised centres.

Over 60% of patients are either under- or overtreated ,

Total cholesterol was lower in males with CAH than in Post Monica. In women, total and HDL cholesterol in CAH was higher than in healthy controls even after adjustment for taking hormonal contraception. We suppose this is due to promoting healthy dietary habits in CAH. However, there was still **high percentage of patients esp. women with significant dyslipidemia**. These results are similar to those of Arlt et al in CAHASE study (JCEM 2010). Blood pressure and glucose levels did not differ from healthy controls, which does not agree with CAHASE (women with classic CAH had higher diastolic pressure , blood glucose was higher than 5,6 mmol/l in 8 % patients)..Bone density was evaluated only in segment of patients, however these preliminary results show **frequent osteopenia/porosis**, which again agree with CAHASE.

Health status of adults with congenital adrenal hyperplasia: a cohort study of 203 patients. Arlt W J Clin Endocrinol Metab. 2010 Nov;95(11):5110-21. (1)