

# Age at diagnosis and mental development in children with congenital hypothyroidism in the absence of newborn screening programme.



Y.Ouarezki<sup>1</sup>, A.Ladjouze<sup>2</sup>, S.Khera<sup>3</sup>, A.Djermane<sup>1</sup>, A.Laraba<sup>2</sup>

<sup>1</sup>Etablissement Public Hospitalier Hassen Badi, El-Harrach, Algiers, Algeria, <sup>2</sup>Teaching Hospital Lamine Debaghine, Algiers, Algeria, <sup>3</sup>Teaching Hospital of Laghouat, Algeria.

## BACKGROUND:

The outcome of congenital hypothyroidism (CH) has dramatically changed since the start of neonatal screening. However the benefit of this program is not felt in Algeria and other countries where the pathology is still causing irreparable brain damage.

## OBJECTIVE AND HYPOTHESES:

To evaluate age at diagnosis of CH and its impact on the mental development in the absence of new-born screening.

## METHOD:

Case notes of all patients referred to our Endocrine Clinic for congenital hypothyroidism (CH) were reviewed. Patients with Down's syndrome were not included in the study. True congenital hypothyroidism was diagnosed when imaging showed thyroid dysgenesis or dysmorphogenesis; or when venous TSH was > 50 mU/L with subnormal fT4 (< 12 pmol/L) in healthy term babies in whom imaging was not performed. Developmental delay was classified as severe, moderate and mild, and analysed according to age at starting treatment (< 1 month, 1-3 months, 3-6 months,>6 months); and to initial venous fT4.

## RESULTS:

Of 75 patients referred with elevated TSH, 56 (F36:M20) had true CH. Mean (range) age at referral was 17.6 (0.4-138) months at which time 21 (37.5%) patients were already on treatment.

True Congenital Hypothyroidism		N=56
<b>Age at referral (months)</b>		
Mean		17.6
Range		0.4-138
<b>Aetiologies</b>		
- Dysgenesis:		29
. Hypoplasia		13
. Athyreosis		12
. Ectopia		4
- Dyshormonogenesis		10
- Unknown cause		16
Mean FT4(pmol/l)		5.9
		0.01-20.5
Mean TSH (mU/l)		132.6
		5.74-1075
Age at starting treatment (months)		10.38
		0.3-138

Table 1: Aetiologies of CH, TFT's and age at starting treatment.

Age at starting treatment (months)	< 1	1-3	3-6	> 6
	N=12	N=20	N=10	N=14
FT4 (pmol/l)	5.8	7.04	5.9	6.1
<b>Mental delay</b>				
- Mild	2	-	-	1
- Moderate	8	7	3	4
- Severe	-	-	4	2

Table 2: Correlation between age at starting treatment, FT4 levels and mental retardation.

Mental delay was considered severe in 6(10.7%) patients, moderate in 22 (39.3%) and mild in 3(5.3%). Severe mental delay was highly correlated with initial ft4 levels:3.83pmol/l (0.99-10.7) and age at starting treatment: 11 (0.5-40) months. The most frequent symptoms were speech delay, attention and concentration deficit, school difficulties. Early psychological support improved mental status of patients with mental deficit.

## CONCLUSION:

Diagnosis of CH is still considerably delayed in countries where newborn screening is absent, leading to mental impairment in affected children. Algerian paediatricians are seriously concerned by this situation and are actively working to set-up a national screening program.

## REFERENCES

- Nguyen Phu Dat et al. Factors affecting on the mental development in children with Congenital Hypothyroidism . Int J Ped End 2013, (Suppl 1):147.
- K.A.Silva et al. Neurodevelopmental Outcomes in Congenital Hypothyroidism: Comparison of Initial T<sub>4</sub> Dose and Time to Reach Target T<sub>4</sub> and TSH. J Ped 2005;147: 775-780
- J.J. Bongers-Schokking et al. Influence of timing and dose of thyroid hormone replacement on development in infants with congenital hypothyroidism. J Ped 2000;136: 292-297.
- D.A. Fisher. The importance of early management in optimizing IQ in infants with congenital hypothyroidism. J Ped 2000;136: 273-274.

