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PP1-229. GROWTH AND SYNDROMES (TO INCLUDE TURNER SYNDROME) 2

"Endocrine evaluation of 29 Cornelia de Lange Syndrome patients (CdLS)"

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INTRODUCTION AND OBJECTIVES

Cornelia de Lange (CdLS) syndrome (OMIM #122470) is a complex disease, characterized by distinctive facial features, failure to thrive, microcephaly, intrauterine growth retardation and anomalies in multiple organ systems. The complexity and severity of the endocrine commitment is variable. *NIPBL, SMC1A, SMC3, RAD21* and *HDAC8* genes, all involved in the cohesin pathway, have been identified to cause CdLS. There are few published studies on the endocrine evaluation in these patients; so our study is aimed at expanding our knowledge in this broad field of research.

METHODS

A descriptive study of 29 Spanish patients diagnosed with CdLS was performed. We have analyzed the different metabolic, anthropometric variables. A complete study was carried out, genetic testing and analyzing different endocrine axes including thyroid, adrenal, growth, gonadal, lipid, phosphocalcic and metabolical study.

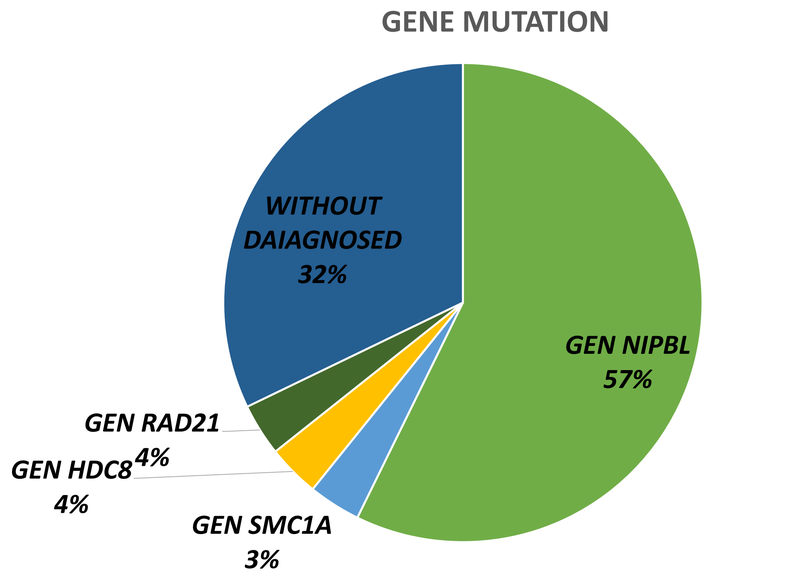
RESULTS

BACKGROUND		TOTAL(n=29)
Gestational		
- **Pregnancy control**	n (%)	29 (100)
- **Gestational diabetes**	n (%)	2 (7,7)
- **IUGR***	n (%)	10 (38,5)
- **Caesarean**	n (%)	9 (33,3)
- **Hypoglycemia**	n (%)	6 (20,7)
- **Admission neonatology**	n (%)	17 (63)
- **Breastfeeding**	n (%)	8 (29,6)
Family		
- **Delayed or early puberty**	n (%)	6 (22,2)
- **Obesity**	n (%)	7 (25,9)
- **Autoinmune disease**	n (%)	15 (55,6)

Table 1. Family and gestational background
*IUGR: Intrauterine growth restriction

PERINATAL HISTORY		TOTAL (n=29)
- Male	n (%)	10 (34,5)
- Premature < 37 SG	n (%)	15 (51,7)
- Birth weight (g)	x̄ ± DE	2371,1±528
- Birth weight W < -2 DE	n (%)	9 (31)
- Birth lenght (cm)	x̄ ± DT	44,4±3,69
- Birth lenght < -2 DE	n (%)	13 (44,8)
- Cranial perimeter (cm)	x̄ ± DT	30,4±1,9

Table 2. Perinatal history of CdlS patients



Graphe 1. Gene mutations of CdlS patientes

ANTHROPOMETRY		TOTAL (n=29)
Weight(Kg)	x̄ ± DE	34,70± 23,90
Weight (SDS)	x̄ ± DE	-1,30 ± 1,80
Height (cm)	x̄ ± DE	123,5 ± 26,40
Height(SDS)	x̄ ± DE	-3,3 ± 1,90
BMI (Kg/m2)	x̄ ± DE	19,99 ± 8,31
BMI (sds)	x̄ ± DE	0,11 ± 2,40
Head circumference (cm)	x̄ ± DE	49,50 ± 4,70
Head circumference (SDS)	x̄ ± DE	-2,92 ± 2,60
Waist circumference (cm)	x̄ ± DE	64,30± 15,60
Waist circumference (SDS)	x̄ ± DE	-0,40 ± 1,70

Table 3. Current anthropometry of CdlS patientes

BIOCHEMISTRY		TOTAL (n=24)
Glycemia (mg/dl)	x̄ ± DE	88,5±10,30
Triglycerides (mg/dl)	x̄ ± DE	123±74,70
Total cholesterol (mg/dl)	x̄ ± DE	164,2±38,7
HDL cholesterol (mg/dl)	x̄ ± DE	45,8±9,20
LDL cholesterol (mg/dl)	x̄ ± DE	93,7±33,60
Insulin (microU/ml)	x̄ ± DE	15,60±9,90
HOMA IR	x̄ ± DE	4,09±4,30
Calcium (mg/dl)	x̄ ± DE	9,62 (0,55)
Phosfores (mg/dl)	x̄ ± DE	3,81 (0,97)
25-0h-vitamin D (ng/ml)	x̄ ± DE	24,30 (5,26)

Table 4. Biochemistry variables of the sample

HORMONAL		TOTAL (n=24)
ACTH (pg/ml)	x̄ ± DE	30,73 (37,37)
Cortisol (mcg/dl)	x̄ ± DE	10,21 (4,47)
Androstendiona (ng/ml)	x̄ ± DE	1,55 (1,00)
17-0H-PG (ng/ml)	x̄ ± DE	0,77 (0,44)
Prolactin (ng/ml)	x̄ ± DE	26,03 (19,80)
TSH (mU/mL)	x̄ ± DE	1,78 (1,12)
T4 Free (ng/dL)	x̄ ± DE	1,22 (0,18)
IGF1 (ng/mL)	x̄ ± DE	173, 68 (83,69)
IGFBP3 (mcg/mL)	x̄ ± DE	4,36 (1,46)
PTH (pg/ml)	x̄ ± DE	40,11 (18,21)
LH (mU/ml)	x̄ ± DE	6,66 (4,64)
FSH (mU/ml)	x̄ ± DE	4,35 (2,95)
Testosterone	x̄ ± DE	0,6 (1,00)

Table 5. Descriptive study hormonal variables

CONCLUSIONS

Our patients have growth retardation, most of them with prenatal onset, according to the clinical criteria of CdLS. Only few reports have commented on endocrine abnormalities in CdLS. In our sample of 29 CdLS patients serious alterations are not observed; however, it is important to carry out an endocrine evaluation, for each individual patient, in order to understand and be able to assess their metabolism adequately.

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