Distal monosomy 10q presented as congenital hypothyroidism

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INTRODUCTION

Distal monosomy 10q - rare chromosomal anomaly.Most 10q deletions occur de novo. Characterized by: slow growth before and after birth, mild to severe intellectual disability and distinctive craniofacial features (hypertelorism, strabismus, a prominent or broad nasal bridge, and posteriorly rotated low-set ears), hyperactivity and impulsivity.

For diagnosis - subtelomeric rearrangements by MLPA test (cytogenetic analysis may not be sensitive enough to detect very small deletions).

CASE PRESENTATION

We report a family (mother and daughter)
Mother - with congenital scoliosis, facial dysmorphism, congenital hypothyroidism with goiter; because the hight doses of Euthyrox® we assumed an enzyme deficiency. She was opereted for goiter and thyroid hormone replacement was made successfully.

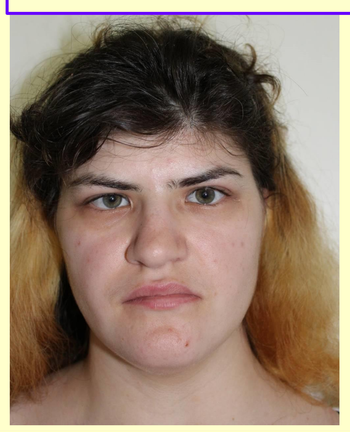
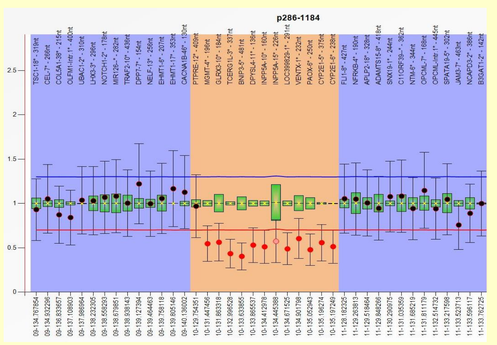


Fig.1 - mother



46,XX.mlpa 10qsubtel (P286)x1

The daugther – the same facial dysmorphism, developmental delay, congenital hypothyroidism (Euthyrox® replacement treatment).

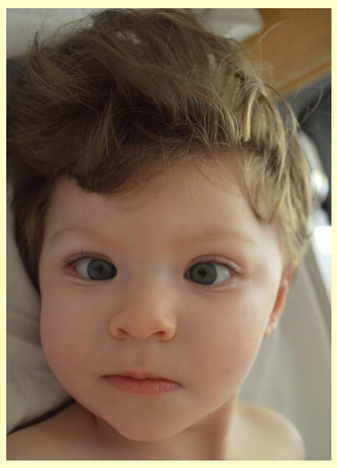
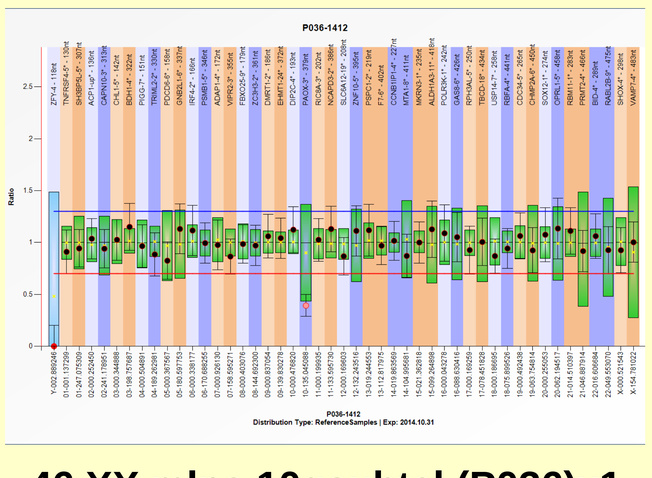
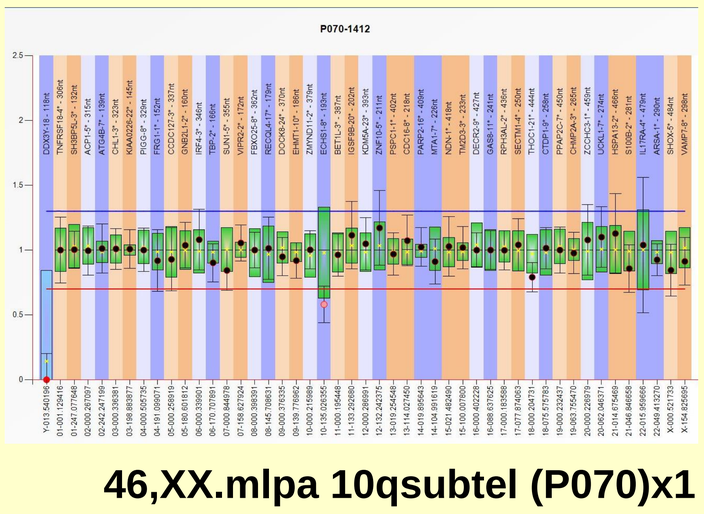


Fig.2 child



46,XX.mlpa 10qsubtel (P036)x1



46,XX.mlpa 10qsubtel (P070)x1

RESULTS

Mother	Child
	Birth history: GA = 37 w, W = 3300g, L = 33 cm, OFC = 31 cm, APGAR = 9 (1')
age 23 y; BMI = 33,98 kg/mp, OFC = -4.15 SD	age 1year 6 months; W = +0.57 SD; H = -3.27 SD; OFC = -1.18 SD, growth velocity = 1.3 cm/month
Facial features: coarse face, broad nasal bridge with a prominent nose, hypertelorism, deep set eyes, strabismus, myopia Early scoliosis (surgical solved), feet – brachydactyly 2-5 Hypothyroidism - congenital	Facial features: broad nasal bridge with a prominent nose, hypertelorism, deep set eyes, epicanthic folds, strabismus, thin upper lip, micrognathia Short neck, congenital torticollis Hand - brachydactyly
Psychomotor retardation – mild (IQ = 42)	Psychomotor retardation
2 months age: **T3 = 0.57 ng/ml ↓** AAT Tg = 20.2 UI/ml (n); AAT TPO = 3 UI/ml (n) 23y age: TSH = 0.786 uUI/ml (n: 0.4-4); fT4 = 1.46 ng/ml (n 0.89-1.76)	TSH = 2.51uUI/ml (n. 0.4-8.4); fT4 = 1.44 ng/dl (n 0.89-1.76) vit D = 50.94 ng/ml (n>30) **serum phosphate = 5,34mg/dl (n = 2.3-4.7)** **alkaline phosphatase = 357 UI/l (n: <200)**
Euthyrox 100 ug/day	**Euthyrox 37,5 ug/day**
MLPA: **46,XX.mlpa 10qsubtel (P286)x1**	**MLPA:** **46,XX.mlpa 10qsubtel (P036)x1** **46,XX.mlpa 10qsubtel (P070)x1**

CONCLUSIONS

The most consistent clinical features in our cases are cranial dysmorphy, growth retardation, developmental delay, hypothyroidism and congenital fingers abnormalities. Management is symptomatic only. In our family the risk of recurrence is theoretical 50%. Once the family chromosome change is known, we recommended prenatal test in any future pregnancy to find out the presence of a subterminal 10q deletion.
The prognosis is unclear as there are no available data concerning long-term follow up.

References

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=96148

http://www.rarechromo.org/information/Chromosome%2010/10q25%20and%2010q26%20deletions%20FTNW.pdf

Tanteles GA, Nikolaou E, Christou Y, Alexandrou A, Evangelidou P, Christophidou-Anastasiadou V, Sismani C, Papacostas SS. Mild Phenotype in a Patient with a De Novo 6.3 Mb Distal Deletion at 10q26.2q26.3. Case Rep Genet. 2015;2015:242891.

Yatsenko SA, Kruer MC, Bader PI, Corzo D, Schuette J, Keegan CE, Nowakowska B, Peacock S, Cai WW, Peiffer DA, Gunderson KL, Ou Z, Chinault AC, Cheung SW. Identification of critical regions for clinical features of distal 10q deletion syndrome. Clin Genet. 2009 Jul;76(1):54-62.

Piccione M, Antona V, Piro E, Cavani S, Malacarne M, Pierluigi M, Corsello G. 10qter deletion: a new case. Am J Med Genet A. 2008 Sep 15;146A(18):2435-8.

Kogasaka R, Morohoshi T, Sawada Y, Fujiwara M. Terminal deletion of chromosome 10q and its clinical features. Acta Paediatr Jpn. 1990 Feb;32(1):83-7.

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Thyroid

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