P-173

Inherited Xp22.2-22.13 Duplication Presenting with Global Developmental Delay: A Case Report

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INTRODUCTION

Xp22.2-22.13 duplication is a rare genetic mutation worldwide. In 2011, Sismani C et al. reported a familial Xp22.2-22.13 duplication (9Mb) from mother and daughter and four affected sons. Only male carriers presented with mental retardation, developmental delay, cardiovascular problems, and mild dysmorphic facial features. However, It is known to have not been previously reported in Korea. Here we present a case of Xp22.2-22.13 duplication (704Kb) with a global development delay.

CASE REPORT

A 26-month-old boy with a developmental delay was referred to our clinic. He had been delivered at 41 weeks of gestation. His maternal second cousin had an intellectual disability. Bayley -III Scales-based developmental evaluation administered at 26 months of age showed his cognition was around 18 months, gross motor skills were around 17 months, fine motor skills were around 20 months, receptive communication was around 14 months, and expressive communication was around 12 months.

Chromosome analysis showed a normal male karyotype. An array comparative genomic hybridization (array CGH) revealed Xp22.2-22.13 duplication (704Kb, 16,997,258-17,701,211), which was also found in the patient's mother.

Brain MRI showed no definite structural abnormality. In diffusion tensor tractography, right optic radiation were not reconstructed compared to the other side. The right frontotemporal segment of arcuate fasciculus showed reduced volume and low fractional anisotropy (FA) value. A low FA value was observed in the right cingulum and inferior fronto-occipital fasciculus (Figure 1) (Table 1).

The follow-up study conducted at 35 months also showed developmental delay (Table 2). On the Developmental Test of Visual–Motor Integration (VMI) at 36 months, his age equivalent was 1 year, 4 months. Serial language assessments (PRES) were conducted until he reached 57 months, and he showed language developmental delay on all tests (Table 2).

Bayley-III Scales	26m 20d		35m 25d	
	DAE	Composite Score	DAE	Composite Score
Cognition	18 months	70	26 months	85
Receptive communication	14 months	62	24 months	77
Expressive communication	12 months		21 months	
Fine motor	20 months	73	23 months	70
Gross motor	17 months		21 months	
Social-emotional	-	70	-	75
Adaptive behavior	-	62	-	70

PRES		26m	33m	38m	51m	57m
Receptive communication	Score	Uncheckable	5	5	14	17
	Quotient				62.7	61.4
Expressive communication	Score	0	1	2	8	12
	Quotient				60	52.6

DAE = Developmental age equivalent, PRES = Preschool receptive-expressive language scale

Table 2. Bayley Ⅲ Scales of Infant Development and PRES (Preschool Receptive-Expressive Language Scale)

Tract	Tract volume (No of fibers)		Mean FA	
	Right	Left	Right	Left
Corticospinal tract	1290	1265	0.6166	0.6097
Cortico-ponto-cerebellar tract	745	874	0.5986	0.6349
Corpus callosum	6011		0.6128	
Uncinate fasciculus	203	194	0.4632	0.4668
Arcuate fasciculus (Fronto-temoral)	254	396	0.4808	0.5105
Arcuate fasciculus (Fronto-parietal)	548	362	0.4886	0.4604
Cingulum	1051	1014	0.4572	0.4995
Inferior fronto-occipital fasciculus	496	362	0.4725	0.5099
Inferior longutidinal fasciculus	736	898	0.5176	05066
Optic radiation	59	279	0.5676	0.5057

Table 1. Tract volume and mean fractional anisotropy (FA) of Diffusion tensor tractography

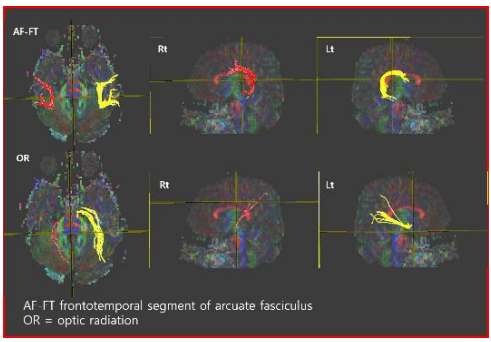

Figure 1. Diffusion tensor tractography

CONCLUSION

We report on a case of inherited Xp22.2-22.13 duplication with global developmental delay and white matter evaluation. Further study is needed to determine the specific role of the genes in this region in the development process.