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Cervical spine malformations with 5q34-5q35.2 micro interstitial deletion: case report

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Introduction

In our report, we identified a proband with a 'de novo' 5q34-q35.2 deletion. The purpose of this report is to present a unique clinical phenotype such as skeletal anomaly in cervical spine and reviewed possible related genotype.

Case report

A 4-years-old girl visited our rehabilitation clinic for development delay. She was born at 39 weeks of gestation with a birth weight of 2,000g. She underwent cardiac septal defect surgery at 2 months and hospitalized for feeding and respiratory disorders for 7 months. On physical examination, she had dysmorphic features with microphthalmos, decreased visual and hearing acuity, so that she applied an artificial eye and hearing aids. Developmental milestones were markedly delayed. Holding up of head were recognized at 12 months, while creeping and crawling at 3 and 4 years respectively, and she could not stand up at 4 years of age. Chromosome 5q34-5q35.2 micro interstitial deletion was diagnosed by Fluorescent in situ Hybridization. Further genetic testing for Prader Willi syndome, Angelman syndrome and spinal muscular atrophy proved negative. A notable symptom was persistent head tilting, which has previously been considered to be associated with symptoms of hypotonia. However, she also had a limitation of range of motion of head rotation. Under suspicion of musculoskeletal disorders, we performed further radiographic analysis. Cervical plain radiographs showed abnormal tube-like structures in posterior C5 [Fig 1] and whole body spine plain radiographs demonstrated thoracolumbar scoliosis [Fig 2]. Non-enhance cervical spine 3 Dimensional Computed Tomography (3DCT) revealed that a bone cleft in the right pars interarticularis, a bone defect in both lamina of C5 and agenesis of the right articular process of C5, which associated with bony fusion of C4-C5. Subluxation in craniocervical joints was also showed in cervical spine 3DCT. An additional focal bone defect in transverse foramen caused inferior bony protrusion and right foramina stenosis at C2-3 [Fig 3]. Bayley Scales at 41 months suggested cognitive, receptive communication, expressive communication, fine motor, and gross motor skill equivalents of 5, 2, 3, 5, and 7 months, respectively. Total score of Gross Motor Function Measure (GMFM) at 41 months was 29% and her GMFCS level was 4. Her speech was limited to simple vocalization without meaning. Despite the intensive rehabilitation therapy, her developmental abilities were significantly delayed at 5 years 10 months of age.

Conclusion

Several deformities of cervical spine in this patient has not been previously described in the 5q deletion encompassing MSX2. MSX2 has been known as involved in the processes

of limb, craniofacial formation. Based on review with 5q related patients, MSX2 gene could be proposed as candidates for skeletal anomaly involving cervical spinal abnormalities. Molecular studies are required to identify the genotype-phenotype correlation of this deletion.

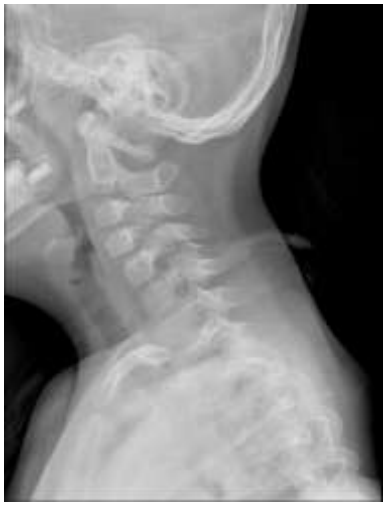

Fig 1. Cervical X-ray showed abnormal tube-like structures in posterior C5.

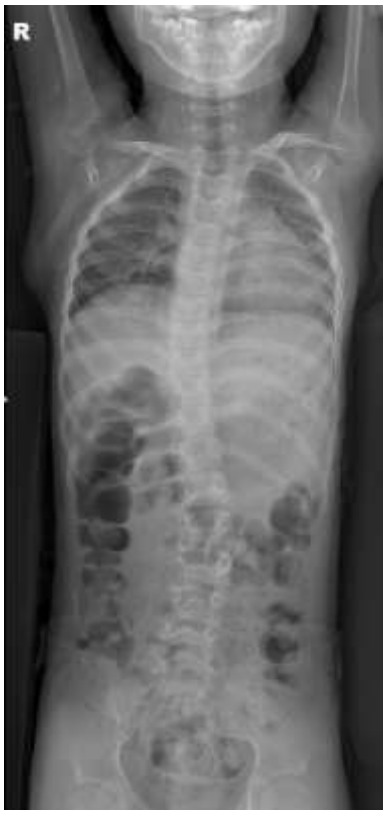

Fig 2. Whole body spine plain radiographs demonstrated thoracolumbar scoliosis.

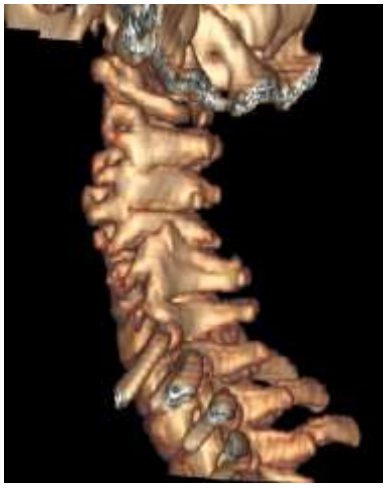

Fig 3. C-spine CT revealed bone defect in both lamina of C5 ,agenesis of the right articular process of C5 with bony fusion of C4-C5.