Clinical Genetics

# AB024. Etiology of recognizable or unclassified overgrowth syndrome

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**Background:** Overgrowth syndromes comprise a group of disorders associated with excessive growth and other features such as facial dysmorphism, developmental delay (DD)/intellectual disability (ID), congenital anomalies, neurological problems and an increased risk of neoplasia. Here, we report 13 children with overgrowth syndrome.

**Methods:** A total of 13 unrelated patients with overgrowth syndrome were recruited. All of them presented with (I) excessive height >95th percentile with/without (II) DD/ID, and at least two minor features of the following: (i) dysmorphic craniofacial features and (ii) congenital anomalies.

**Results:** Seven patients were diagnosed as Sotos syndrome (SS) whish was confirmed by sequencing or MLPA analysis of *NSD1* gene. Five patients had Beckwith-Wiedemann syndrome (BWS) confirmed by methylation PCR. The other patient was found to have interstitial microdeletion of 7q22.1-7q22.3 by array comparative genomic hybridization. All SS patients showed macrocephaly, tall stature, and DD/ID. Among seven patients with SS, 3/7 (42.8%) patients had seizure and thoracolumbar scoliosis; 3/7 (42.8%) patients had attention deficit hyperactivity disorder (ADHD); 1/7 (14.3%) patient was found to have periventricular leukomalacia (PVL) as shown on brain MRI. Among patients with BWS, 4/5 (80%) patients had DD/ID; 3/5 (60%) patients showed hemihypertrophy which is left-side dominant; 2/5 (40%) had umbilical hernia and history of neonatal hypoglycemia; 1/5 was complicated with hepatoblastoma; 1/5 reported intractable seizure requiring antiepileptic medications. Clinical manifestations of the patient with 7q22.1-7q22.3 microdeletion included generalized excessive growth, prominent forehead, mild hypertelorism, undescended/retractile testis, and global DD. The deletion was starting from nucleotide 102,877,293 extending to nucleotide 105,121,326 which involved 9 genes.

**Conclusions:** Careful clinical examination of patients may allow the delineation of clinically recognizable overgrowth conditions. A detailed molecular analysis of the rearranged regions may provide the clues for identification of the genes involved in growth regulation.

**Keywords:** Overgrowth syndrome; molecular analysis; growth regulation

doi: 10.21037/atm.2017.s024

**Cite this abstract as:** Kun C, Kim YM. Etiology of recognizable or unclassified overgrowth syndrome. Ann Transl Med 2017;5(Suppl 2):AB024. doi: 10.21037/atm.2017.s024

 