Lumbosacral spina bifida in Pallister-Killian syndrome: A case report

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INTRODUCTION
Pallister-Killian syndrome (PKS) is a rare disorder caused by tissue limited mosaic tetrasomy of 12p. Prenatal findings include diaphragmatic hernia, rhizomelic upper limbs, nuchal edema, internal abnormalities, omphalocele and fetal hidrops (2,3). It is diagnosed with cytogenetic analysis performed from amniocytes, chorionic villus, fetal blood or fibroblast culture. Lymphocytes are usually cytogenetically normal (4).

CASE REPORT
A pregnant woman at 19-week gestational age showed high risk for neural tube defect in second trimester screening. Omphalocele (O) and irregularity in vertebral alignment (V) were noted in fetal ultrasonography evaluation. Additionally, fetal echocardiography showed left ventricular hypoplasia (LVH). Cytogenetic analysis was not performed in chorionic villus or amniocytes. The pregnancy was terminated at 20-week gestational age on demand of the parents.

POSTMORTEM FETAL EXAMINATION AND FIBROBLAST CULTURE
Omphalocele (O), sacral appendage (SA) and lumbosacral spina bifida (LSB) were observed at postmortem examination. Also note the coarse and flattened appearance of the face, depressed nasal bridge, short neck, brachydactyly and dorsal pedal edema. PKS was suspected and fibroblast culture was done.

RESULTS AND DISCUSSION
Cytogenetic analysis revealed non-mosaic 47,XY,i(12)(p10) karyotype and confirmed the diagnosis. This is the first case of PKS with lumbosacral spina bifida. Cytogenetic analysis is the mostly used method to diagnose PKS and differentiate it from Fryns syndrome which may bear common clinical findings (5).

References