

## Makale

Short Communication

### Anti-N-Methyl-D-Aspartate Receptor Encephalitis that Developed after Herpes Encephalitis: A Case Report and Literature Review

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Neuropediatrics

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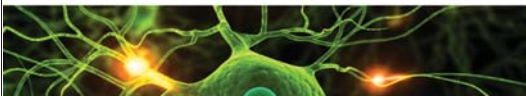
Özel İmmünoloji Birimimizin, Ankara Üniversitesi Tıp Fakültesi Pediatrik Nöroloji, Pediatrik Enfeksiyon ve Radyoloji Bölümleriyle ortaklaşa sunduğu vaka takdimi, "Anti-N-Methyl-d-Aspartate Receptor Encephalitis that Developed after Herpes Encephalitis: A Case Report and Literature Review" başlığı ile *Neuropediatrics* Dergisinde yayımlandı.

## Sunumlar

**NÖRONAL ANTİKORLAR**

Dr. Tutku Taşkınoğlu  
Düzen Laboratuvarlar Grubu

07 Haziran 2014



Özel İmmünoloji Birim Sorumlumuz Uz. Dr. Tutku Taşkınoğlu, Gazi ÜTF Mikrobiyoloji Anabilim Dalı tarafından düzenlenen IFA (*Immunofluorescence Assay*) Kursu kapsamında "Nöronal Antikorlar", Klinik Mikrobiyoloji Uzmanlık Derneği (KLİMUD) tarafından düzenlenen IFA Kursu kapsamında "İnflamatuvar Bağırsak Hastalıkları ve Çölyak Tanı/Takibinde Laboratuvar" kurslarını vermiş ve 4-7 Haziran 2014 tarihlerinde Ankara'da düzenlenen 8. Ulusal Moleküler ve Tanısal Mikrobiyoloji Kongresi'nde de "Nöronal Antikorlar" başlıklı sunumu yapmıştır.

Mikrobiyoloji Birim Sorumlumuz Uz. Uğur Çiftçi de, 18-19 Eylül tarihlerinde Nevşehir'de düzenlenen Sağlık Bakanlığı

Türkiye Halk Sağlığı Kurumu Başkanlığı - Bulaşıcı Hastalıkların Laboratuvara Dayalı Sürveyansı Sempozyumu kapsamında "Mikrobiyoloji Laboratuvarında Kalite Kontrolün Önemi ve Dış Kalite Değerlendirme Çalışmaları" başlıklı sunumu yapmıştır.

## Posterler

- 17-20 Mayıs 2014 tarihleri arasında Boston, ABD'de düzenlenen *114<sup>th</sup> General Meeting of the American Society for Microbiology* (ASM 2014) Kongresinde, Mikrobiyoloji Birimimiz "Klinik açıdan önemli mikroorganizmaların identifikasyonunda iki farklı MALDI-TOF MS sisteminin konvensiyonel fenotipleme veya 16S rDNA bazlı identifikasyon ile karşılaştırılması" başlıklı posterini sunmuştur.
- 31 Mayıs-3 Haziran 2014 tarihleri arasında Milano, İtalya'da düzenlenen European Human Genetics Conference kapsamında, Moleküler Genetik Birimimizin katkılarıyla hazırlanmış olan "*Leri Weill syndrome findings in an infertile man with 45,X/46,X,derY,t(Y;Y)(p11.2;q11.21) and SHOXY deletion*" ve "*PIK3CA mutations in Non-Small Cell Lung Cancer*" başlıklı posterler sunulmuştur.

**Leri Weill syndrome findings in an infertile man with 45,X/46,X,derY,t(Y;Y)(p11.2;q11.21) and SHOXY deletion**

Yararbaş K1, Sağlam-Ada B2, Alp F2, Çavdarı B3, Ocal A2, Gürkan S1, Karatag G1, Akın I1, Bilecen K1, Laleli-Sahin E1, Tükun A1

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**INTRODUCTION**

Leri-Weill dyschondrosteosis (LWD) (OMIM# 127300) or syndrome is a rare genetic disorder with skeletal dysplasia characterized by short stature, mesomelia, and Madelung deformity. We report a 29-year-old male with Leri Weill Syndrome phenotype which is caused by 45,X/46,X,derY,t(Y;Y)(p11.2;q11.21) and SHOXY deletion.

**CASE REPORT**

29-year-old male was referred from Urology department because of azoospermia. Infertility was the first complaint that forced him request medical Simultaneously regular consultations, radiographic and other procedures and genetic analysis were planned. (Please refer to Table 1 for OMIM Clinical Synopsis. Features of the syndrome and comparison with

Parameter	Reference	Present Case
Sex	Male	Male
Age	29	29
Stature	158 cm	158 cm
Hand/foot	Small	Small
Skull	Normal	Normal
Spine	Normal	Normal
Wrist	Normal	Normal
Genetics	Normal	45,X/46,X,derY,t(Y;Y)(p11.2;q11.21) and SHOXY deletion

**Radiographic features** Madelung deformity, flattening of the vertebral spine were revealed by X Ray axiometer survey (Figure 2).

**Cytogenetics, FISH and Array CGH**

Conventional cytogenetic analysis and Y chromosomal microdeletion scan were performed. No microdeletions were present on the loci 5Y14(SRY), 5Y84, 5Y86 (AZF), 5Y127, 5Y134 (AZFb), 5Y254, 5Y255 (AZF), 5Y132, 5Y153 (AZFc) and 5Y151. Chromosomal analysis revealed a mosaic finding of two series: 45,X and 46,X with a derY, possibly t(Y;Y)(p11.2;q11.21). Additional metaphase FISH analysis with a commercial probe targeting DDX1, LSI SRY supported the mosaic numerical finding with 15% of the metaphases only showing one DDX1 signal without LSI SRY and 45 chromosomal material on DAPI stain. Whole chromosome painting FISH studies demonstrated that the derived Y chromosome consisted of only Y chromosomal material. SHOXY analysis by FISH technique revealed absence of SHOXY signal but 2 DY2 signals on derived Y chromosome. Finally, chromosomal microarray findings (Affymetrix CytoScan 750K SNP Array platform) confirmed a SHOXY deletion with no additional significant findings above cut-off values.

**DISCUSSION**

Leri-Weill dyschondrosteosis/syndrome (LWD/LWS) is a skeletal dysplasia first described in 1929. The classic clinical triad of LWS is short stature, mesomelia and Madelung deformity. The inheritance of this disorder follows an pseudo-autosomal dominant pattern. Penetrance of the phenotype appears to be incomplete within families. Besides, females are more frequently affected than males and clinical features, such as bilateral Madelung deformity and short stature, have been described as being more severe in females than in males. LWS is caused by heterozygous defects in the short stature homeobox-containing gene (SHOX) or by deletion of the SHOX downstream regulatory domain. The SHOX gene is located in the telomeric pseudoautosomal 1 region (PAR1) on the short arm of both sex chromosomes and escapes X inactivation. 2,4,5 SHOX is expressed on both sex chromosomes in males and females and there is no difference between SHOX (X) and SHOX (Y) and that's why SNP array platform revealed a deleted SHOX on "Y" (Figure 3). Our patient is a male Leri Weill Syndrome caused by haploinsufficiency of SHOX gene due to 45,X and 45,X/46,X,derY,t(Y;Y)(p11.2;q11.21) and SHOXY deletion. He is macrocephalic and his vertebral spines are exaggerated flattened which are not reported in clinical synopsis of LWS. Duplication of Y q11.21 and deletion of Yp11.21 may be responsible these phenotypes or additional undetected defects might be present in the patient.

Please note that parental karyotype is not available, but the cytogenetic defect could be considered as de novo and mosaicism as a postzygotic mitotic error. We suggest that azoospermia in our patient is more likely to be the result of an inability of the X and Y chromosomes to pair normally during meiosis due to recombination of Y chromosome and deletion of pseudoautosomal region on Yp.

**References**

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