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MEETING ABSTRACTS

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The CGH-Array was normal in both parents and brother, but the patient presented a deletion of 3,04 Mb in 4q31.21 and a duplication of 212,03 Kb in 9q34.2 (arr[hg19] 4q31.21(143,272,775-146,310,106)x1,9q34.2(137,103,263-137,315,288)x3 Abnormal Male). Both duplication and deletion are "de novo" and not previously described. The 4q31.21 deletion includes 14 genes (11 OMIM). The 9q34.2 duplication includes the RXRA OMIM gene, without triplo-sensitivity previously described.
The pathologic potential of chromosome aberrations detected "de novo" can be difficult to determine, and it is necesary to have the phenotype of the patient in mind as well as the genes involved in the deletion/duplication. In our case we believe that both detected chromosomal alterations, alone or in conjunction, are responsible for the patient's phenotype.
Consent to publish: The authors confirm that written informed consent was received by the patient for publication.

1.P79

A rare case report of Langer-Giedion syndrome/ Trichorhinophalangeal syndrome (TRPS) type II and Cornelia de Lange syndrome 4

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This abstract is not included here as it has already been published.

Tumour Cytogenetics

2.P1

WAGR region deletions size and position do mean

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Introduction
WAGR syndrome (OMIM#194072) is caused by deletions of 11p13 encompassing the PAX6 and WT1 genes. Almost all cases are sporadic. Four WAGR classical clinical signs rare present in complete combination. The 1.6 Mb region is supposed to be critical for all classical WAGR criteria, though clinical diagnosis is based on identified 11p13 deletion, aniridia in combination with at least one other feature.
Aims
Study of association between clinical criteria of WAGR syndrome and 11p13 deletion length and position.
Methods
MLPA-, FISH.
Results
14 Russian patients with WAGR region deletions, 4 males and 10 females from 2 months to 40 years old, were studied. 12 cases were sporadic while 1 was familial. Unilateral nephroblastoma developed in 6 patients within 2 first years of life, 4 adult patients had no tumor, 4 kids under 3 years old had not developed it (probably yet). 3 out of 6 patients with WAGR syndrome and the largest deletions (>7.5 Mb) showed unilateral nephroblastoma and severe neurological deficit combined with mental retardation. The rest of the patients with congenital aniridia or WAGR syndrome associated with narrower deletions demonstrated milder cognitive and neurologic status. Unilateral nephroblastoma developed in 1 out of 5 patients carrying the narrowest deletions with the same right breakpoint at the WT1 level (0.7–2.2 Mb). 4 patients under 3 years old are still in the high oncological risk group despite the lack of tumor, as they possess gross deletions of critical region. They require rigorous monitoring.
Conclusion
Risk for Wilms' tumor development in patients with 11p13 deletion in a cohort of Russian patients is 43% – 71%. Prognosis for the Wilms' tumor development and cognitive deficit worsens with deletion region increase towards centromere from WT1 locus.
The research was partially supported by grants RFFR 17-04-00288; 15-04-01859, RSF 17-15-01051.

2.P2

The importance of conventional cytogenetic analysis in monitoring the progression of haematological malignancies

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Aims
To highlight the importance of cytogenetic investigations in detecting disease progression after cancer treatment.
Methods
Four leukemic patients were treated but subsequently developed new haematological malignancies. A variable number of tandem repeat test was run to correlate the degree of transplant engraftment for patients who did a bone marrow transplant (BMT).
Results
Case 1: A female patient in 2012 diagnosed with Acute Lymphoblastic Leukaemia (ALL) had a t(9;22)(q34;q11.2) rearrangement. She underwent a BMT in 2013 and remained in remission until 2015. Despite VNTR showing 100% donor engraftment, cytogenetics revealed that her bone marrow (BM) had undergone transformation to Myeloproliferative Disorder.
Case 2: A female patient with ALL in 1995 underwent a BMT and was in remission with 100% donor cells. In 2015, a t(12;18) rearrangement detected in her bone marrow. This was not seen previously, suggesting either disease progression or donor cell constitutional rearrangement. Donor karyotype was recommended to rule out constitutional rearrangement but was not forthcoming.
Case 3: A female patient with Acute Myeloid Leukaemia in 1995 had a complex karyotype. She had a BMT in 1997 and was in remission until a relapse in 2007. A second BMT in 2009 showed 100% donor engraftment. In 2014, cytogenetics detected a completely different karyotype, suggesting malignant transformation.
Case 4: A male patient admitted in 2013 for Severe Aplastic Anaemia and suspected MDS had a complex karyotype. He had a sex-mismatched BMT in 2014. His BM never reached 100% engraftment as the karyotype showed persistence of the malignant clone and normal donor cells. Three months later, cytogenetics detected disease progression.
Conclusion
BMT is the only cure for haematological malignancies. Routine karyotyping is necessary to detect malignant transformation. This could alter the patient treatment. The study shows that 100% donor engraftment does not necessarily mean 100% treatment success.