2,500 g. The examination findings were: small frontal and occipital asymmetry, upslanted palpebral fissures, hypertelorism, flat nasal bridge, board neck, bilateral clinodactyly, mild mental retardation and small penis (both testes were in well-formed scrotal sacs). No cardiovascular alterations were detected.

Chromosome culture and karyotyping were performed using standard techniques and showed a karyotype of 49,XXXXY.

Although initially 49,XXXXY pentasomy was considered a variant of Klinefelter syndrome, it is currently recognized as a separate clinical entity distinguished by facial features, multiple skeletal and cardiac defects, and short stature. A 49,XXXXY karyotype is thought to arise from maternal non-disjunction which occurs during both meiosis I and meiosis II. This produces a secondary oocyte with four X chromosomes, which, when fertilized by a Y chromosome-bearing sperm, results in an embryo with 49,XXXXY syndrome.

In this case, the facial dysmorphism and small penis were the main features which led to a suspicion of sex chromosome aneuploidy that was confirmed by chromosomal analysis.

The prognosis of these children depends on the extent of severity of the condition, while the management mandates a multidisciplinary approach with pediatric endocrinology, pediatric surgery, orthopedics, psychiatry and clinical genetics evaluation.

Keywords: Small penis, Sex chromosome aneuploidy, Facial dysmorphism, XXXXY syndrome

1.P30

Large interstitial del(13)(q13q14.3): the importance of detailed clinical information in cytogenetic studies

Fernanda Paula Oliveira[1], Natália Oliva Teles[1], Joana Ribeiro[1], Manuela Mota Freitas[1], Margarida Azevedo[2], Hildeberto Correia[1], Maria da Luz Fonseca e Silva[1]

[1]Centro de Genética Médica Jacinto Magalhães, INSA Departamento de Genética—Unidade de Citogenética, Porto, Portugal

[2]Centro Hospitalar Entre Douro e Vouga Serviço de Pediatria, Santa Maria da Feira, Portugal

may accompany the deletion, including mental retardation and craniofacial dysmorphism. The severity of the phenotype depends on the extent of the deletion. Retinoblastoma is a malignant tumor in the retina and is the most common ocular cancer in children. The association of most cases of retinoblastoma with an interstitial del(13q) has led to the localization of the retinoblastoma gene in 13q14.

We report a case of a boy aged 8 referred for cytogenetic studies, presenting with mild mental retardation, craniofacial dysmorphism, delayed intrauterine growth (IUGR) and retinoblastoma. The karyotype was obtained from peripheral blood lymphocyte cultures using high-resolution GTG banding and standard techniques. Fluorescence in situ hybridization was performed using the LSI 13 (RB1) probe (Vysis) for region 13q14 spanning the RB1 gene.

The chromosomal analysis revealed a large interstitial deletion of the long arm of chromosome 13. Although the exact breakpoints were difficult to establish, the deleted region did not appear to encompass the band which includes the retinoblastoma gene. Molecular cytogenetic techniques showed that the retinoblastoma gene was deleted. This confirmed the clinical indication of retinoblastoma and defined the deletion breakpoints more precisely. Final karyotype: 46,XY,del(13)(q13q14.3).ish del(13)(q14.1q14.3)(RB1−).

Except for the presence of IUGR, the clinical description of this patient is in agreement with other reports in the literature. We would like to emphasize the importance of detailed clinical information that, together with classical and molecular cytogenetic techniques, could be useful in better defining the breakpoints, establishing correct genotype/phenotype correlation and thus providing appropriate genetic counselling. The blood samples of the parents were requested for karyotype analysis in order to clarify this chromosome deletion.

Keywords: del(13q), Interstitial deletion, Retinoblastoma

1.P31

Variable phenotypes in a group of 6 patients with distal deletions of chromosome 9p

Drahuše Novotná, Zdena Vlčková, Jana Drábová, Zuzana Zmítková, Miroslava Hančárová, Markéta Havlovicová, Marcela Malíková, Alena Puchmajerová, Monika Koudová, Zdeněk Sedláček
Charles University 2nd Faculty of Medicine and University Hospital Motol Department of Biology and Medical Genetics, Prague, Czech Republic

Monosomy of the distal part of 9p is associated with a known syndrome characterised by mental retardation (MR), delayed speech development, trigonocephaly, midface hypoplasia, long philtrum and other dysmorphic features. In addition, there exists a large cohort of patients suffering from gonadal dysgenesis with or without MR to stigmatisation. These patients have male karyotypes and deletions of variable sizes involving band 9p24.3. Three genes responsible for sex reversal—DMRT1, DMRT2 and DMRT3, located in 9p24—are missing in patients with gonadal dysgenesis, except one patient described in 2009 by Barbaro et al. Several hypotheses have been proposed to explain this, e.g. that the haploinsufficiency of the DMRT genes could cause gonadal dysgenesis through a dosage threshold effect or that autosomal copy number variants play different roles depending on the XX or XY constitutions.

We present two groups of patients whose karyotypes were determined using G banding and fluorescence in situ hybridization, and, in some cases, also array CGH. The first group includes three girls with female karyotype and derivative chromosome 9 arising in two cases from translocation between 2p and 9p, and in one case from translocation between 9p and 12q. All three girls present with features of the monosomy 9p syndrome, but their phenotypes could also be influenced by partial trisomies of the other chromosomes. The second group consists of three patients with male karyotype and pure distal 9p deletions. The external genitalia and sexual development of the first boy are normal, but he has MR, autism and dysmorphic features. The second child is a baby with ambiguous genitalia. The third patient is a young girl with amenorrhea but apparently normal phenotype without MR, although her deletion is the largest within this group. Our observations confirm the complexity of the genotype–phenotype correlation in carriers of distal 9p deletions.

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Keywords: Deletion of 9p, Monosomy 9p syndrome, Mental retardation, Gonadal dysgenesis

1.P32

A rare inherited case of 4q deletion detected by GTG and array analyses in a newborn

Manuela Ariza Cobos[1], Luz Miguez[1], Francisco Barros[1], Victoria Pardo[1], Cristina Lois[1], Raquel Martínez[2], Manuel Macía[3], Fernando Domínguez[1]
[1]Fundación Pública Galega de Medicina Xenómica, Prenatal Diagnoses, Santiago de Compostela, Spain
[2]Hospital Cristal Paediatrics, Orense, Spain
[3]Hospital Clínico Universitario de Santiago de Compostela, Gynecology, Santiago de Compostela, Spain

Terminal 4q deletions are rarely described (<100 cases). Most of them are de novo with simple deletions of a small chromosome segment. Only a few cases are inherited from parental carriers of balanced translocation or unbalanced abnormalities (14% of the cases with 4q deletion).

We present a case of an inherited 4q32.1qter deletion and 10q15pter trisomy detected by GTG and array analyses in a newborn.

The proband was referred at the age of 25 days for genetic evaluation with 46,XY,?der(4)t(4;?)(q2?5;?) karyotype. Clinical features are hypotonia, cardiovascular defects (such as interauricular communication, ductus arteriousus), cryptorchidism, a cleft in the palate, clinodactyly, inguinal hernia and bilateral hydronephrosis.

Cytogenetic studies were performed in the parents and revealed a maternal balanced translocation, 46,XX,t(4;10)(q32;p15). Our case result from an inherited unbalanced segregation to genome-wide screening using array analyses (Cytogenetics whole-genome 2.7M array, Affymetrix) showed terminal 36-Mb 4q deletion, mapped between 155130472 and 191125000 positions and 1.51-Mb duplication located at 10p15, mapped between 0 and 1238648 positions. Final karyotype was corrected as follows: 46,XY,der(4)t(4;10)(q32.1;p15).

We compared the clinical symptoms of our patient with those of other cases of 4q32 monosomy reported in the literature in order to establish an accurate phenotype–genotype correlation and to provide genetic counselling to these families.

Keywords: 4q deletion syndrome, Array analyses