line mosaicism in an asymptomatic father of a boy with cardiac malformations and a microdeletion of 22q11. Methods: Chromosome analysis and FISH was performed on chromosome slides from the DiGeorge patient, his parents and his grandparents using routine GTG-banding and FISH probes LSI TUPLE1 SpO/LSI ARSA SpG and alpha XT680. Results: The father showed the karyotype 46,XY,del(22) (q11q11)[65%]/47,idem,+inv dup(22) (pter-q11::q11-pter) [12%]/47,idem,+r(22)[23%]de novo. The inverted duplication and the ring chromosome both included the 22q11 region. The son inherited the 46,XY,del(22)(q11q11) cell line. The karyotypes of the mother and the grandparents were normal. Conclusion: We hypothesize that the del(22) in the father evolved through the primary formation of an inv dup(22). The latter was rearranged into a ring chromosome which was subsequently lost during cell division, thereby giving rise to three different cell lines. The recurrence risk of DiGeorge syndrome was estimated to 30–50%, in addition there is also a theoretical risk of offspring with cat eye syndrome.

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Detection of de novo partial trisomy of 3p with FISH

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We report a case of partial trisomy 3 in a seven month old girl with exophthalmus, small nose, microgynathia, hypertelorism, hepatosplenomegaly, hypotonia without infant reflexious. The chromosome preparation obtained from peripheral blood lymphocytes revealed an abnormal 4p chromosome. The karyotype was 46,XX,der(4)(p15). Parental karyotypes were normal. Fluoresence in situ hybridization (FISH) was performed using chromosome painting probe 4 and multicolor probe. FISH analysis revealed the presence of chromosomal part of 3p. Thus, the karyotype was reinterpreted as 46,XX,der (4)(p15).ish dup (3)(p25)(wpc3+). Further step for this case, molecular genetic study for her parents was planned to identify the parental origin of the partial trisomy of 3p. The findings of clinical and molecular genetic will be discussed with the related literature.

P 585

Ataxia teleangiectasia mutated (ATM) gene deletion in Non-Hodgkin's lymphoma: correlation with clinicobiological features

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Ataxia-teleangiectasia (AT) is an autosomal recessive multy-sistem disorder that derives from the inheritance of a structurally abnormal gene at 11q22.3-q23.1 referred to as ataxia-teleangiectasia mutated (ATM) gene. A-T subjects present chromosomal instability and a strong predisposition to develop lymphoid tumors, especially T-cell prolymphocytic leukemia. In order to ascertain the clinicobiologic significance of acquired ataxia-teleangiectasia gene deletion (ATM-) cytogenetic and fluorescence in situ hybridization (FISH) analyses were performed in 135 non-Hodgkin's lymphomas (NHL). An hemizygous ATM- was seen in 44–88% of the cells in 18 cases (13.3%): 5 patients had a B-cell high grade NHL, 8 patients had B-cell low-grade NHL, 5 patients had a T-cell lymphoma. Twelve out of 18 ATM-patients had a complex karyotype, 13 out of 18 had more than 90% abnormal metaphases. Patients with ATM- had more frequently a T-cell phenotype ($p = 0.009$), a complex karyotype ($p = 0.001$), as compared with patients without ATM-. Dual color cohybridization of a BCL1 probe or of a TCL1 probe and of the ATM probe showed ATM- to be a secondary event in 4 BCL1 rearranged cases and to be an early event in 2 TCL 1 rearranged lymphomas. A highly significant correlation was found between ATM- and shorter survival ($p = 0.0002$). This cytogenetic lesion maintained its prognostic importance in multivariate analysis ($p = 0.0036$), along with complex karyotype ($p = 0.0171$), performance status ($p = 0.0005$). serum LDH level ($p = 0.0055$), male sex ($p = 0.0010$) and splenomegaly ($p = 0.0268$). Though not representing a primary genetic lesion in the majority of cases acquired ATM- has clinicobiologic importance in NHL, possibly representing a major determinant of outcome.

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A case of circular chromosome 11 – r(11) (p15; q23)

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Some cases of describing circular chromosome 11 with points of breaking in the terminal parts of chromosome 11 have been known. It has been marked that the clinical picture of circular chromosome 11 is connected with the loss of part 11 q23*qter. The proband, a nine year old mentally retarded boy was timely born with the body mass 2,600 g. From the third pregnancy. His mother was 21 by the time of the boy's birth, his father was 24. His two elder brothers are normal. During his first year the boy was retarded both psychomotorically and physically. At examination his body mass was 16.5 kg, height 109 cm. The diagnosis is oligophreny in the grade of debility. Objectively: trigonocefalus, hypertelorism, mongoloid shape of eyes, wide back of the nose, high palate, "carp's" mouth, micrognatia, vision pathology (longsightedness, squint), short neck, thoracic scoliosis. No pathology in inner organs. The child's karyotype is 46,xy, r(11) (p15; q23). The mother's karyotype is normal. The father and brothers have not been examined.

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A case of partial trisomy on chromosome 9 resulting from the mother's balanced translocation (5;9)(p15;q22)

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There exist dozens of descriptions of some cases of trisomy 9 in medical literature. Three forms are distinguished: full, mosaic and the whole length trisomy, excluding segment of long shoulder 9 q32-32*qter. Our case is of particular interest since it presents trisomy of the whole chromosome excluding segment q22*qter. The proband is a three – year old male born from the first 39 week long pregnancy, with the body mass of 2500 g. And length of 49 cm. The pregnancy, was complicated by toxemia and anemia. By the time of the boy's birth his mother was 19 and his father was 23. Examining the boy at the age of 3 months the following anomalies of development were found out: hydrocephalus (cysts of the brain), soft cranial bones, wide cranial seams, flat occiput, a small forehead, hypotelorism, epicapt, deformed low-placed ears, microstomia, a gotic palate, a short neck, infundibuliform thorax of II–III grade inborn pidgeon toes, transverse hand fissure, omphaloceli, inguinal hernia, hypoplasia of penis. As for the internal organs: an in born cardiac defect (defect of the interventricle partition), left pulmonary emphysema. The proband's cytogenetic examination revealed trisomy 9 p ter* q22 (an additional chromosome which is a derivative of chromosome 9) resulting from the mother's segregation 3:1 of reciproctal translocation t(5;9)(p15; q22). The proband's karyotype is 47,xy+der(9) mat. The father's karyotype is normal.

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