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Background. The blastic crisis of chronic myeloid leukemia (CML-BC) is usually the final phase of the disease, in which the percentage of the young, often undifferentiated cells, known as blastocytes gets above 20%. Nowadays, in the era of the therapy with Tyrosine Kinase Inhibitors, the transformation from CML to CML-BC occurs much later and more rarely.
Case report. We present the case of a 71 year old male, admitted in July 2017, to the Haematology Unit of Mures County Emergency Hospital presenting severe anemia, leukocytosis, leukocyte left shift, absence of the leukemic hiatus and thrombocytopenia. Splenomegaly (7 - 9 cm) was also found. Cytogenetic examination revealed the presence of Philadelphia chromosome and real-time PCR showed 87% positivity for BCR-ABL. Chronic Myeloid Leukemia was the diagnosis and treatment with Dasatinib was initiated. A month after the patient develops severe thrombocytopenia and hemorrhagic purpura. Treatment was interrupted until the platelet count was restored and continued after with smaller dosage. Erythrocyte mass was transfused in order to correct the anemia. Three months after the diagnosis with CML, spleen expansion and hyperleukocytosis was observed. The peripheral blood smear indicated high blastocyte percentage (88%) and the patient was admitted and diagnosed with CML-BC. The diagnosis was confirmed, RT-PCR still showed positivity for BCR-ABL in 48%. Induction treatment for Acut Lymphoblastic Leukemia with adapted protocol for elderly patient with comorbidities was initiated. In December 2017 the patient refuses further treatment and unfortunately passes away.
Conclusion. Chronic myeloid leukemia is a condition with a high survival rate, especially after introducing the tyrosine kinase inhibitors, but when the blastic transformation occurs, many patients are lost due to infections and hemorrhagic complications.
Key words: myeloid, leukemia, lymphoblastic, tyrosine, kinase.

32. PARTIAL 13 MONOSOMY WITH CORPUS CALLOSUM AGENESIS AND OTHER CONGENITAL ABNORMALITIES – A CASE REPORT

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Background. The corpus callosum comprises the largest tract of nerve fibres in the human brain. It is developed from the telencephalon starting in the 11th week of foetal life. Partial or complete agenesis of the corpus callosum is a rare developmental anomaly of unknown cause. A case of corpus callosum agenesis is described.
Case report. The patient was a small for gestational age (1950g) female infant delivered at 35 weeks. In view of multiple congenital abnormalities (bilateral choanal atresia, atrial septal defect, ventricular septal defect and facial dysmorphism), chromosome studies were done and showed partial monosomy of chromosome 13 (46,XX, del (13)(q22q33)). Head ultrasound and cranial CT scan was performed which found appearances typical of agenesis of the corpus with ascension of the third ventricle and increased distance between lateral ventricles, cerebellar hemispheres and vermis atrophy, cisterna magna and fourth ventricle dilatation. After the surgical intervention for bilateral choanal atresia, a cranial ultrasound was performed and confirmed the atresia of the corpus callosum, but the path of anterior cerebral artery showed on

color Doppler suggests the existence of the rostrum. The patient was carefully monitored after the surgery. 24 days after the surgery the general status of the patient becomes altered, the patient presenting jet vomiting and nystagmus. The transfontanellar ultrasound showed ventriculomegaly with intracranial pressure (IR: 0.79->0.95) and the lumbar puncture showed transparent, sterile cerebrospinal fluid for which she remains carefully monitored.
Conclusions. Although rare, agenesis of the corpus callosum is easely recognisable on CT scan and neonatal ultrasound. Even it is itself asymptomatic, may be associated with other malformations, especially in genetic syndromes, playing an important role in the production of neurological symptoms.
Key words: congenital malformations, genetic syndrome, corpus callosum agenesis

33. MYELOID PROLIFERATION ASSOCIATED WITH DOWN SYNDROME: A CASE REPORT

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Background. Atypical chronic myeloid leukemia (aCML), BCR-ABL1 negative is a rare myelodysplastic syndrome (MDS)/myeloproliferative neoplasm (MPN) for which no current standard of care exists. ACML is characterized by many clinical features (splenomegaly, myeloid predominance in the bone marrow with some dysplastic features but without a differentiation block) and laboratory abnormalities (myeloid proliferation, low leukocyte alkaline phosphate values). A review of the literature suggests that the presence of an abnormal chromosome 21 may predispose to the development of leukemia.
Case report. A 41-year-old man with a past medical history of Down syndrome (47, XY, +21) was admitted to the Haematology Unit of Mures Country Emergency Hospital with severe anemia, thrombocytopenia and leukocytosis. Following the peripheral smear, bone marrow biopsy and RT-PCR for bcr/abl (negative) indicated atypical chronic myeloid leukemia or myelodysplastic syndrome(MDS)/myeloproliferative neoplasm(MPN) grade III. The substitution treatment was established and the condition of the patient has evolved unfavorably with bronchopneumonia, respiratory failure, diffuse micropapous rash and Clostridium colitis. Due to chromosomal abnormality the cytostatic treatment is difficult to administer. The RT-PCR for JAK2, cMPL, CALR was negative. The final diagnosis falls as a myeloid neoplasia associated with Down syndrome with blasts lower than 20% at the medullary level, but in terms of WHO classification, the blastic percentage is not relevant. Therefore, the treatment chosen was mild cytoreduction (ARA-C) and substitution depending on tolerance, but even with the correctly administered treatment the patient died after ten months.
Conclusions. In conclusion, atypical chronic myeloid leukemia is a rare disease and the association with chromosomal abnormalities and the lack of standards of care is a challenge in treating these patients and poor results should be expected.
Key words: Down syndrome, myeloid proliferation

34. MYOCLONUS-DYSTONIA MASQUERADING AS WILSON

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