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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