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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Background. Myoclonus-dystonia is a movement disorder that typically affects the neck, torso, and arms. Individuals with this condition experience quick, involuntary muscle jerks or twitches (myoclonus). About half of individuals with myoclonus-dystonia develop dystonia, which is involuntary tensing of various muscles that causes unusual positioning. In myoclonus-dystonia, dystonia often affects one or both hands, causing writer's cramp, or the neck, causing the head to turn (torticollis).
Case report. A 26-year-old man consulted his doctor because of involuntary movement of the eyeball and involuntary muscular contractions on the left side of the body. It progressed to involuntary movement of the neck (torticollis) and left arm. Postural tremor is also present in the left arm. The laboratory results showed slighty diminished ceruloplasmin enzyme and blood copper, but the urine copper was three times higher than normal. The patient has been suspected of Wilson's disease but the genetic test came out negative and treatment with cuprinil has been ineffective. The patient also tried taking Isicom (levodopa), Romparkin (central anticholinergic), Haloperidol (neuroleptic) with no benefit. In 2014 he suffered a surgery for herniated disc C5-C6 with no symptom improvement. The cerebral MRI and electromyography has shown no significant changes. The Wilson disease diagnosis can be excluded, but to confirm Myoclonus dystonia, it's needed to make another genetic molecular testing for SGCE. This gene translates a transmembrane protein in the dystrophin associated glycoprotein complex found in skeletal muscle and neurons. If the test is positive, the most effective treatment would be a surgical intervention of Deep Brain Stimulation (DBS) in internal globus pallidus and the central intermediate nucleus of the thalamus, which can cure both the myoclonus and dystonia.
Conclusions. This case illustrates the difficulty of assigning a clear diagnosis regarding dystonia and the complexity of etiologies. Nonetheless, a correct diagnosis and treatment can improve the patient's life quality substantially.
Key words: myoclonic-dystonia, torticollis, Wilson's disease, DBS

35. SPONTANEOUS PNEUMOTHORAX: CONGENITAL CYSTIC ADENOMATOID MALFORMATION OF THE LUNG

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Background. Congenital cystic adenomatoid malformation (CCAM) is a rare developmental abnormality of the lung with an incidence of 1 in 25,000 to 1 in 35,000 pregnancies and represents 25% of congenital lung malformations and 95% of congenital lung lesions, males and females being equally affected. It is characterized by multicystic mass arising in the lung parenchyma from abnormal proliferation of the immature terminal respiratory bronchioles. The clinical spectrum is wide and ranges from silent incidental lesions to severe congenital respiratory distress or stillbirths. The diagnosis of CCAM can be made in utero by prenatal ultrasongraphy and postnatally by imaging radiography and CT. We report a case of a CCAM, which provoked spontaneous pneumothorax.
Case report. Eight month old patient was delivered to the Emergency Department at Institution of Mother and Child with pneumonia and left spontaneous pneumothorax. From the anamnesis, after birth was discovered CCAM after which the patient took a conservative treatment. On examination the patient was agitated, presented acrocyanosis, the general condition of the patient was severe. Chest X-ray showed a large left-sided pneumothorax with mediastinal shift to right and air-containing cystic area of left lower lobe. Pneumothorax was drained immediately, drain tube was seen through left 5th intercostal space. CT scan control of the chest showed expanded left lung with multiple cystic lesion in the lower lobe. After pre-operative preparation lower left