receptor mutation is related with a severe intracellular microorganism infection. We describe an 11 years old boy with partial IFNγ-receptor deficiency, diagnosed at 7 years old in a context of *Mycobacterium* intracelullare multifocal osteomyelitis, medicated with antibacillar (9 months) and IFNγ immunotherapy (2 years). One year after suspending IFNγ-therapy, he was admitted with unilateral, painful red eye, blurred vision, photophobia, and tearing complains for the last 15 days, cough and chest pain within 1 month and lower limbs nummular rash. Conjunctival exam reveals 360-degree perilimbal injection, endothelium keratitic precipitates and visual acuity decreased in the affected eye. Results of laboratory investigations including urine, blood and skin cultures, *Mycobacterium-avium* complex DNA detection and viral serologies were negative except for high levels of *Mycoplasma pneumoniae* IgM antibodies. The uveitis subsided with topical application of steroids, cycloplegia, and oral azithromycin, kept as profilactic therapy three times weekly. IFNγ-therapy was reinitiated improving patient condition. The authors emphasize the exuberant *Mycoplasma* infection, in a patient with IFNγ receptor deficiency after suspending IFNγ-therapy, so far not described. *Mycoplasma*, as an intracellular agent, may play an important role in infection of patients with impaired Interferon γ-mediated immunity.

1257
Ataxia-telangiectasia - review of five cases

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Background: Ataxia-telangiectasia (A-T) is a primary immunodeficiency disease, characterized by neurological problems (ataxia), ocular and skin telangiectasias, recurrent respiratory infections and predisposition to neoplastic disease. It is a hereditary disease (autosomal recessive), caused by mutation in the *ATM* gene on chromosome 11.
Case reports: We report five cases (3 male, 2 female) of patients with A-T, currently aged between 6 and 17 years old (median: 11 years). Ataxia became evident between 2 and 3 years of age, and the diagnosis of A-T was made between 3 and 6 years of age. In all patients there was progressive neurological deterioration, with onset of ocular apraxia between ages 3 and 6, loss of ability to walk independently between 7 and 13 years (requiring to use a wheelchair) in three cases, and the presence of ocular and skin telangiectasias. The most relevant clinical features are the frequent respiratory infections (three patients), the episodes of diarrhea associated with Giardia lamblia infection (two patients) and the decreased growth rate. On analytical test results, 4 patients have increased alpha-fetoprotein levels and low lymphocyte count, mainly in the T CD4+ subpopulation. There are IgA deficiency (4 patients), low levels of IgG (1 patient), and low or absent IgE (five patients). The karyotype presents structural abnormalities involving chromosomes 7 and 14, and an increased chromosomal instability. The radiosensitivity assay (done in two patients) showed increased chromosomal breakage after exposure of cell cultures to ionizing radiation. Analysis of mutations in the *ATM* gene is ongoing.
Comments: A-T is a rare hereditary disease with progressive, irreversible neurological deterioration and immunodeficiency. It has a poor prognosis, with pulmonary infections and cancer being the leading causes of death. An early diagnosis can improve and prolong the quality of life of these patients and allows for genetic counselling. Patients with a mutated gene also have an increased incidence of cancer, so vigilance is mandatory.

1258
Successful treatment of pemphigus foliaceus with cyclosphosphamide: a case report

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Background: Pemphigus foliaceus is a rare multifactorial autoimmune disease characterized by transient cutaneous blisters in a seborrheic distribution; it may appear at any age. Acantholysis in the upper epidermis is appreciated histologically. The autoimmune nature of pemphigus was used as rationale for the use of immunosuppressive drugs and plasmapheresis.
Case report: We report a case of severe pemphigus foliaceus successfully treated with cyclosphosphamide. A 70 year-old male was admitted to our Division showing severe desquamative erythema on his face and chest. He referred an initial onset of eczematous manifestations into the armpit 5 years before, regressed by steroid treatment. During the following years the patient developed diabetes mellitus, chronic renal failure, chronic bronchitis and hypertensive heart disease. He came to our observance because of a reappearance of cutaneous lesions, with vasculitic, exfoliative, urticarial, scratching characteristics and accompanied by itching and burning.He was red-faced and the desquamation involved all the face. A punch biopsy confirmed the diagnosis. In the meanwhile, laboratory investigations were launched. Glycemia and renal function indexes were altered compatibly with the concomitant diseases. Complement and Ig dosage, coagulation indexes and the other routine exams were normal. The researched autoantibodies (ANA, anti-ENA, AMA, ASMA, anti-nDNA, ANCA, anticardiolipin, antiendomysial and antitransglutaminase) had not a significative title or were absent. Underway infections were excluded. HSV IgG and VZV IgG have been instead found present. No altered levels of tumoral markers (CEA, CA 19.9, CA 125, TPA) have been observed. Immunophenotyping on peripheral blood highlighted an increase of CD56 lymphocytes and a lower rate of CD19 cells. The initial therapeutic measures included prednisone 15 mg/die, doxiciclin 100 mg/die, rupatadine 10 mg/die, i.v. immunoglobulins (15 mg), without improvement. So a treatment with intravenous and then oral cyclosphosphamide, combined with steroids (methylprednisolone 20 mg/die i.v. for 3 days and then prednisone 10 mg/die), was undertaken. Signs and symptoms progressively reduced. At the checks during the subsequent month, the patient maintains the response to the immunosuppressive drug, without collateral effects.
Conclusions: We reported this case as example of successful combined use of systemic steroid and cyclosphosphamide in treatment of the pemphigus foliaceus.

1259
Natural killer cell deficiencies in children with herpetic encephalitis - a case series

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NK cells play a fundamental role in innate and in early phases of adaptive immunity against viral infections, both in humans and in animal models. Their role in immunity against herpes viruses is well established too, and NK cell deficiencies have been correlated with severe herpetic infections, as documented by single case reports. We investigated a series of children consecutively admitted to the Paediatric Hospital of our University with diagnosis of herpetic infection and CNS involvement.

Our data show that numeric and/or functional deficits of NK cell populations are present in everyone of the five patients: flow cytometric analysis showed that in three of these children (case no. 1, 2 and 4) circulating NK cells and CD56+ T cells were considerably decreased, while the other two children (case no. 3 and 5) resulted to be carrier of CD16 gene polymorphisms associated with NK functional immunodeficiency, in heterozygosis and homozygosis, respectively. No significant deficits of other branches of immunity were detected in all but one of them (case no.2), who had absolute lymphocytopenia. Further investigations, involving functional assays, are ongoing.

These findings emphasize the role of NK-related immunodeficiencies in determining increased incidence and worse outcome of viral infections in humans, in accord with previously published single case reports. This is of much greater importance if we consider that even if the first report of a child with severe herpetic infection and NK deficiency dates back to 1989, NK cell populations are still rarely investigated in these conditions. Despite the lack of an actual therapeutic approach, being able to identify higher risk patients would allow avoiding voluntary exposure to infected contacts, prompt instauration of anti-viral therapies in case of infection and higher degree of attention in those conditions, such as pregnancy or concomitant use of immunosuppressive drugs, in which risk of severe injury is elevated.

1260
Acute hemorrhagic edema of childhood: an usual variant of leucocytoclastic vasculitis

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Infantile acute hemorrhagic edema (IAHE) is a rare leucocytoclastic vasculitis that affects children younger than 2 years of age. It is described as a unique clinical entity although some clinicians postulate that IAHE is a variant of infantile Henoch-Schonlein purpura limited to the skin. The clinical presentation of IAHE is impressive because of the sudden appearance and spread of the purpuric rash and when it is accompanied by fever resembles often meningococcemia, a primary differential diagnosis. But the clinical hallmark of IAHE is the good condition of the child and the benign outcome, usually with no complications or reccurence.

Case presentation: We report a case of a 12-month old girl with clinical and histopathologic findings that are typical of infantile acute hemorrhagic edema. A 12-month old girl presented at our clinic with inflammatory edema of the lower limbs and purpuric lesions with acute onset developing less than 12 h. The physical examination showed a good clinical condition and the child was afebrile. There were edematous violet-colored, nummular lesions in a target-like format on the limbs and on the face. The rest of the physical examination showed no systematic involvement. The histopathologic examination showed leucocytoclastic vasculitis. After 10 days the lesions disappeared spontaneously and there was no relapse.
Conclusion: The above support the notion that IAHE is a benign disorder, despite the dramatic appearance and with clinical features that justify its characterization as a unique disorder distinct from Henoch-Schonlein purpura.

1261
Catastrophic antiphospholipid syndrome occurring postpartum in a young female

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Background: We present a case of Catastrophic Antiphospholipid Syndrome (CAPS), a life-threatening, multi-organ thrombosis occurring acutely.
Results: This is a case of a 31 year-old female, G2P1 (1001), diagnosed with hyperthyroidism 2 years prior, given methimazole for 1 month, rendering her euthyroid. FT3 was low on her 9th week of this pregnancy, which normalized during her 24th week without any medications. Post-caesarean delivery, she developed sudden hypotension, oliguria, dyspnea, tachypnea and bradycardia. Laboratory findings showed thrombocytopenia, elevated liver enzymes, prolonged activated partial thromboplastin time, elevated D-dimer and Troponin-I. On echocardiography, multiseptal wall motion abnormality and ejection fraction of 30% were seen. CAPS was entertained, and she was treated with enoxaparin, hydrocortisone, and intravenous immunoglobulin with dramatic improvement.
Conclusion: This is a rare case of CAPS, which is a multiorgan thrombosis that has a 50% mortality rate, successfully managed with anticoagulation, steroids, and IVIG.

1262
A novel familial Th2-type autoaggressive granulomatous disease with life-threatening large vessel vasculitis

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A 54 year old patient with combined exokrine and endocrine pancreatic insufficiency presented in reduced general condition showing pancreatic atrophy and retroperitoneal lymphadenopathy on CT scan. Echocardiography revealed a distended aortic root (6.9 cm) including the aortic valve necessitating prosthetic replacement. Histology showed a severe granulomatous inflammation of the perivascular tissues and a similar picture in the enlarged aortoilical lymph nodes. There was no evidence for tuberculosis or sarcoidosis and a broad autoantibody screening was negative. Besides a mild eosinophilia (0.8 G/l, $n < 0.4$) no major hematologic changes were found. Serum protein electrophoresis revealed a polyclonal gammopathy (IgG/IgA) and an increased total IgE (1238 kU/l, $n < 70$). Seven and 5 years before, biopsies of unclear swellings of the parotid gland and the left lower lid could be attributed to an epitheloid granulomatous inflammation as well and an idiopathic thrombocytopenia was successfully treated with corticosteroids 10 years before. A similar clinical and histological picture, but with associated destructive polyarthritis, was found in one sister of the patient. In both patients, leukocyte phenotyping showed a striking Th2-polarization with more than 30% CRTH2-positive T-helper cells. These Th2-memory cells produced, apart from IL-4, IL-5 and IL-13, high amounts of TNF-alpha, in contrast to mouse Th2 cells which rather produce IL-10. Furthermore, basophils showed aberrant spontaneous activation and IL-4 secretion. A corticosteroid therapy was initiated, but already three months later a spontanous fracture of the left femur (suspected bone involvement) necessitated an intensified therapy. We chose a monoclonal anti-TNF alpha antibody (infliximab 5 mg/kg) because of its known antigranulomatous effect and the cytokine profile mentioned above. MR-angiography showed no signs of large vessel vasculitis after three infusions and therapy was continued.
Conclusion: To our knowlegde, this is the first description of a Th2-type granulomatous disease with familial background. One may assume that the histological changes were even present years before diagnosis albeit not necessarily in childhood. Genetic