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"PERSONA: FOR A NEW INCLUSIVE CITIZENSHIP" MUNA FOR SCIENCE, PEACE, EDUCATION: RETHINKING MEDITERRANEAN PROJECT

Claudio Pignata, Francesca Galgano, Università di Napoli Federico II, MUNA

INTRODUZIONI ALLA SESSIONE "MEDITERRANEAN PRIMARY IMMUNODEFICIENCY NETWORK (MEDIPINET) FOR INBORN ERRORS OF IMMUNITY"

C. Pignata, Dipartimento di Scienze Mediche Traslazionali (DISMET) - Sezione di Pediatria, Università di Napoli Federico II, session coordinator.

M.R. Barbuche, Department of Immunology, Institut Pasteur de Tunis

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The Tunisian experience of Severe Combined Immunodeficiency

I. Ben Mustapha, Department of Immunology, Institut Pasteur de Tunis

Diagnosis and treatment of Inborn Errors of Immunity: Clinical, immunological, and molecular characteristics of typical and atypical Severe combined immunodeficiency - a single-center experience over a period of 13years (2008–2021)

B. Belaid, Department of Medical Immunology, Beni-Messous University Hospital Center

Clinical and immunological features of 96 Moroccan children with Severe Combined Immunodeficiency phenotype: Two decades' experience

A. Bousfiha, Laboratory of Clinical Immunology, Inflammation, and Allergy, Faculty of Medicine and Pharmacy of Casablanca, King Hassan II University; Clinical Immunology Unit, Department of Pediatric Infectious Diseases, Children's Hospital, Centre Hospitalo-Universitaire Averroes

Egyptian Experience with Severe Combined Immunodeficiency: 10-year experience

A. Elmarsafy, Pediatrics department, Faculty of Medicine, Cairo University

Severe combined immunodeficiency in Palestine

A. NaserEddin, An-Najah National University, Nablus, Palestine, H Clinic Specialty Hospital

Proposal of a comparative study on Severe Combined Immunodeficiencies among Countries of the Mediterranean area

G. Giardino, I. Ben Mustapha, R. Barbouche, C. Pignata, Dipartimento di Scienze Mediche Traslazionali (DISMET) - Sezione di Pediatria, Università di Napoli Federico II, Department of Immunology, Institut Pasteur de Tunis

INTRODUZIONI ALLA SESSIONE "CULTURE OF DIFFERENCES, HUMAN RIGHTS IN THE MEDITERRANEAN BASIN"

F. Galgano, Dipartimento di Giurisprudenza, Università di Napoli Federico II, session coordinator; C.V. Giosafatto, Dipartimento di Scienze Chimiche, Università di Napoli Federico II

CONTRIBUTI

Palestinian Women in Education and Science

M. Abdalrazeq, Faculty of Medicine and Health Sciences, An-Najah National University

Linguistic education of Syrian refugees in Turkey:the role of "Amal for Education" on the border between two identities

S. Orselli, Università per Stranieri di Siena

ABSTRACTS

THE TUNISIAN EXPERIENCE OF SEVERE COMBINED IMMUNODEFICIENCY

I. Ben Mustapha, Department of Immunology, Institut Pasteur de Tunis

Severe combined immunodeficiency (SCID) is the most severe form of Inborn Errors of Immunity (IEI), characterized by complete absence of T cell-mediated immunity, resulting in a broad-spectrum susceptibility to multiple pathogens. There is considerable genetic heterogeneity, as 17 different conditions all resulting in a SCID have been fully characterized.

SCID accounts approximately for 10% of all diagnosed Tunisian patients with IEI. One hundred and thirty-three patients were diagnosed between 1993 and 2021, including 56 T-B+ SCID patients, 44 T-B-SCID patients and 33 patients with Omenn syndrome.

Genetic screening of the following candidate genes (RAG1, RAG2, ARTEMIS, ADA, IL2RG, JAK3 andIL7RA) were performed in only 23 patients and allowed the identification of IL2RG deficiency in 8

patients, IL7RA deficiency in 3 patients, RAG 2 deficiency in 3 patients, PNP deficiency in one patient and RAG1 deficiency in 8 patients.

Despite the high rate of consanguinity in Tunisia favoring autosomal recessive forms, X-linked inheritance remains well represented among Tunisian SCID patients. The use of NGS will be critical, in the North-African context, to rapidly deliver a molecular diagnosis in order to offer an early diagnosis and an appropriate curative and preventive approach.

DIAGNOSIS AND TREATMENT OF INBORN ERRORS OF IMMUNITY: CLINICAL, IMMUNOLOGICAL, AND MOLECULAR CHARACTERISTICS OF TYPICAL AND ATYPICAL SEVERE COMBINED IMMUNODEFICIENCY - A SINGLE-CENTER EXPERIENCE OVER A PERIOD OF 13 YEARS (2008–2021)

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Background: Severe Combined Immune Deficiency (SCID) is an inherited defect in lymphocyte development and function that results in life-threatening opportunistic infections in early infancy. The age of presentation is variable but occurs typically between 3 and 6 months when the protective effect of maternally transmitted immunoglobulin has diminished, although atypical and late presentations are well described. Data on SCID from developing countries are scarce.

Objective: To describe clinical, laboratory features, and molecular features of SCID and its variants diagnosed at single center in Algiers, Algeria.

Methods: we conducted a 13-year retro-prospective single center study. We collated clinical, laboratory, and molecular details of 169 patients with clinical and immunological profile suggestive of typical or atypical SCID based on 2018 ESID working definition for SCID and classified according to IUIS criteria.

Results: We obtained data on 177 children; 89 were categorized as SCID (57 T-B-NK+, 14 T-B+NK-, 7 T-B+NK+, 11 T-B-NK-), 25 as Omenn syndrome, and 63 as atypical SCID (55 TlowBlowNK+; 5 TlowB+NK+; 3IKBKB). Male-female ratio was 104:73. Median age of onset of clinical symptoms and diagnosis was 3 months (1-5) and 5,5 months (3-11,25), respectively. Molecular diagnosis was obtained in 32 patients. Only 2 children received hematopoietic stem cell transplantation (HSCT). Mortality was recorded in 118 children (66%).

Conclusion: We emphasize that the number of diagnosed patients with SCID reported in this study does not reflect the actual numbers of these disorders in Algeria, because most of the patients with severe forms die in their early life before their diagnosis can be made. Early diagnosis and treatment (HSCT) of these conditions are critical to minimizing mortality and improving quality of life.

CLINICAL AND IMMUNOLOGICAL FEATURES OF 96 MOROCCAN CHILDREN WITH SEVERE COMBINED IMMUNODEFICIENCY PHENOTYPE: TWO DECADES' EXPERIENCE

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Severe combined immunodeficiency (SCID) is a heterogeneous group of primary immuno deficiency diseases (PIDs) characterized by a lack of autologous T lymphocytes. This severe PID is rare, but has a higher prevalence in populations with high rates of consanguinity. The epidemiological, clinical, and immunological features of SCIDs in Moroccan patients have never been reported. The aim of this study was to provide a clinical and immunological description of SCID in Morocco and to assess changes in the care of SCID patients over time.

This cross-sectional retrospective study included 99 Moroccan patients referred to the national PID reference center at Casablanca Children's Hospital for SCID over two decades, from 1998 to 2020. The case definition for this study was: age < 2 years, with a clinical phenotype suggestive of SCID, and lymphopenia, with very low numbers of autologous T cells, according to the IUIS Inborn Errors ofImmunity classification.

Our sample included 50 male patients, and 66% of the patients were born to consanguineous parents. The median age at onset and diagnosis were 3.3 and 6.5 months, respectively. The clinical manifestations commonly observed in these patients were recurrent respiratory tract infection (82%), chronic diarrhea (69%), oral candidiasis (61%) and failure to thrive (65%). The distribution of SCID phenotypes was as follows: T−B−NK+ in 44.5%, T−B−NK− in 32%, T−B+NK− in 18.5%, and T−B+NK+ in 5%. An Omenn syndrome phenotype was observed in 15 patients. SCID was fatal in 84% in the patients in our cohort, due to the difficulties involved in obtaining urgent access to hematopoietic stem cell transplantation, which, nevertheless, saved 16% of the patients.

The autosomal recessive forms of the clinical and immunological phenotypes of SCID, including the T-B-NK+ phenotype in particular, were more frequent than in Western countries. A marked improvement in the early detection of SCID cases over the last decade was noted. Despite recent progress in SCID diagnosis, additional efforts are required for genetic confirmation and particularly for HSCT.

EGYPTIAN EXPERIENCE WITH SEVERE COMBINED IMMUNODEFICIENCY: 10-YEAR EXPERIENCE

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SCID is the commonest encountered PID category among Egyptian children, comprising 30.2% of all diagnosed PID cases in our cohort. Two hundred-thirty SCID patients were diagnosed between 2011 and 2021. Ninety-six (42%) were females and 134 (58%) were males. One hundred-forty patients were diagnosed with T-B-SCID, seventy-nine with T-B+SCID and eleven with Omenn Syndrome.

Forty-three patients had RAG1/RAG2 deficiency, nine had ADA deficiency, thirteen patients had JAK3 deficiency, eight had IL2RG deficiency, four had IL-7RA deficiency and DCLERIC was identified in 4 patients. In three patients with T-B-NK+ phenotype with microcephaly, NHEJ1 variant was detected in 2 siblings and LIG4 in one patient. CD247, CD3E and LAT were identified in one patient each.

In addition, 22 patients were diagnosed as MHC-II deficiency using FCM. Those patients presented with variable clinical and immunological phenotypes. Unlike the European society for Immunodeficiency (ESID) criteria for diagnosis of MHC-II deficiency, most of them had lymphopenia with low T and B cells counts. Nine samples for patients diagnosed with MHCII deficiency were confirmed by whole exome sequencing and revealed variants in RFXANK in 5 patients from 4 families, RFX5 in 3 patients and CIITA in 1 patient.

Among some patients with isolated CD4 lymphopenia and variable serum immunoglobulin levels with markedly reduced recent thymic emigrants (CD4+CD45RO+CD31+), genetic testing revealed RAG1 mutations in 2 atypical patients, DOCK2 mutations in 3 and CARD11 pathogenic variants in another 2 patients. PNP variants were identified in 5 patients from 3 different families having neurological impairment associated with progressive decrease in T cell counts. Other genetic variants were identified for CID patients in Zap70, CD40L and ARPC1Bin 1 patient each.

Prenatal diagnosis was offered for the detection of affected fetuses in families with known mutations in RAG1 in 16 pregnancies, RAG2 in 6 pregnancies and ADA in one pregnancy.

Fourteen SCID/CID were treated with hematopoietic stem cell transplantation (HSCT), with a favorable outcome in 9 patients (8 from matched related siblings) and the loss of five patients including two following a haploidentical HSCT. One ADA-SCID patient was successfully treated with gene therapy.

SEVERE COMBINED IMMUNODEFICIENCY IN PALESTINE

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Primary immunodeficiency diseases (PIDs) are rare life-threatening inborn disorders of the human immune system, characterized by defects of the immune system predisposing individuals to variety of manifestations, including recurrent infections, autoimmunity, malignancy and unusual vaccine complications.
Early recognition of clinical, immunological, and genetic features in patients with PID especially SCID may lead to timely therapeutic intervention with allogeneic stem cell transplantation (SCT).

In the Palestinian population a high rate of consanguinity, up to 40%, favors a high prevalence of recessive form of rare genetic diseases including PIDs.
In Palestine, we are talking about population of around 5 million in the west bank and Gaza strip, during the last decade we diagnosed many cases of SCID and profound combined immunodeficiency (pCID) including new disease-causing genes, all of this with collaboration with other countries, and still we have many new patients and patients without definite diagnosis "leaky SCID", and we estimate that the incidence rate of SCID is much higher than the international incidence rate with autosomal recessive SCID being more common including RAG1 and 2 and JAK3.

The diagnosis of these forms of immunodeficiency requires a close collaboration of experienced clinicians, laboratory immunologists and geneticists in order to diagnose known forms of pCID and SCID and discover new causes. This collaboration will also aid inphysician and nurse education, geneticcounseling, increased awareness of the risks of consanguineous marriage, and the implementation of neonatal screening for the identification of infants with SCID and this will contribute and help indecreasing the incidence of PID especially SCID in the Palestinian population and preventing vaccine complications especially BCG vaccine which are given to all newborns at age of one week.

The discovery of new genetic defects underlying combined immunodeficiency will benefit the affected patient and his family, because access to accurate diagnosis will allow informed medical advice regarding further treatment strategies and genetic counselling.

PROPOSAL OF A COMPARATIVE STUDY ON SEVERE COMBINED IMMUNODEFICIENCIES AMONG COUNTRIES OF THE MEDITERRANEAN AREA

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Inborn errors of immunity (IEI) are rare congenital disorders affecting the development and/or the function of the immune system. The most severe form is SCID (severe combined immunodeficiency) characterized by early onset life threatening infections that usually cause death in the 1st year of life if not promptly recognized and treated. SCID may be due to alterations of different genes and may be transmitted in 2 different modalities, namely autosomal recessive and X-linked. The autosomal recessive modality is more common in population where there is a higher consanguinity rate. Recent studies suggest that the incidence of SCID is largely underestimated and that many patients die before receiving the diagnosis. We recently evaluated clinical, immunological and molecular features of SCID patients followed up at centers of the Italian Network for Primary Immunodeficiency (IPINet) since1986. Interestingly 22.5% of the population were foreigners and were coming from Countries with higher consanguinity rate. The prevalence of SCID in our cohort was significantly higher in the last decade of observation compared to the first 2 decades. However, the estimated prevalence in the last decade of observation was 1:100000 born alive and it was significantly lower compared to the incidence of SCID evaluated through the newborn screening program in the United States. We also observed a different distribution of cases in different Italian Regions with a higher number of cases in regions with specialized centers, suggesting that the presence of a specialized center may improve the diagnostic process and reduce the underestimation. In several cases, we observed a significant diagnostic delay, and this was associated with a worsening of the clinical conditions. In conclusion our data showed that immunological expertise and newborn may reduce the underestimation of the diagnosis and diagnostic delay, improving the management of SCID. The comparison of our experience with that of other Mediterranean countries may help identifying problems that hinder good medical care for IEI paving the way to develop efforts to improve the diagnosis and the management of IEI in the Mediterranean area.

PALESTINIAN WOMEN IN EDUCATION AND SCIENCE

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Science is crucial for the elevation of any society and for its development. Both men and women should work side by side to achieve this progress and development. Through the last decades, women took a major role in scientific life even though in many societies and countries women's life in science was full of difficulties and limitations.

In Palestine, you can find millions of little stories, none less inspiring than the other, in which many Palestinian women showed intelligence skills and the ability to learn and teach science in a

stressfulenvironment [1], those women were strong enough to fight against the occupation from seventy years ago until now and also against the bad traditions to change society thoughts, thus, they were able to gain their rights in life and science. Women brought up under occupation are known to have immense strength. Some had to hold their lifeless sons or brothers in their arms, some were brought up in refugee camps and all struggled to sleep at night with explosions shaking their walls. Certainly, women have made great progress in recent years and achieved a better standard of education: female literacy rose from just 35 % in 1990 to nearly 50 % in 2000. Over the same period, however, male literacy improved, from 63.5 % to 71 %. In the academic year 2010/2011, girls accounted for around half of all students enrolled at Palestinian schools, making up 49.5 % of the primary school population and 54.2 % of secondary school students [2].
Last year's statistics show an even stronger female presence in higher education, where 57.2 % of students were young women. Among graduates, too, women form a clear majority–around 60 % in the academic year 2008/2009 [2].

Finally, as Palestinian women we believe that science is the way to improve ourselves and our whole life, we still need more communications and international collaborations to improve the quality of research and education that we need, and that's what we are looking for.
References:

1. 8 Powerful Palestinian Women You Should Know-Scoop Empire. Access on 26 of January 2022.
2. Palestinian women are well educated, but do not find good jobs | D+C-Development + Cooperation (dandc.eu). Access on 26 of January 2022.

LINGUISTIC EDUCATION OF SYRIAN REFUGEES IN TURKEY:THE ROLE OF "AMAL FOR EDUCATION" ON THE BORDER BETWEEN TWO IDENTITIES

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This research is the result of an experience carried out in the city of Kilis, Turkey, by the non-profit organisation "Amal for Education" in the field of language education of Syrian school-age refugees. It was carried out as part of the degree course in Linguistic Sciences and Intercultural Communication at the University for Foreigners of Siena for the purpose of preparing a master's thesis.
Turkey has been characterised during many centuries by long and varied migrations across its borders, and today is the country with the highest number of refugees and displaced persons in the world. Out of the total of 4 million registered refugees, 91% are of Syrian origin. Since the outbreak of the civil war in Syria in 2011, Turkey has adopted an 'open door' policy towards people fleeing the war, with no restrictions on the number of entries until the final closure of its borders in 2015, due to the intensification of the conflict and the growing threats to Turkish border towns.

Today there are 3.7 million Syrian refugees in Turkey and the reception policies adopted by the country have been (and continue to be) crucial, although not always timely, in responding to the humanitarian emergency. At the beginning, reception policies were intended to host Syrian refugees for a short time, as the most widespread opinion was that the conflict in Syria would be resolved soon. However, the continuation of the Syrian difficult situation meant that the emergency turned to ordinariness, as regards especially Turkish authorities, and the main consequence was the readjustment of the aforementioned reception policies to include Syrians in Turkish society in all respects.

The education of Syrian refugees in Turkey is one of the pivotal points within the reception policies that the Turkish government has adopted and modified during the years of the emergency. The turning point was in 2016 with the gradual closure of the Temporary Education Centres (TECs), present both inside and outside the refugee camps, which until then had provided millions of school-age Syrians with the opportunity to receive an Arabic language education without interrupting the learning process due to war. With the gradual inclusion of Syrians into the Turkish school system, however, the Arabic component of the education has been excluded in favour of an exclusive use of Turkish, depriving Syrian children of the opportunity to continue learning in their own language.

This is the context for the field work of the non-profit association "Amal for Education", which has been working in Kilis since 2013 on language education for Syrian refugees in Turkey. The educational idea behind the association's activities is based on the recovery of linguistic skills. In particular, the affective, identity and cultural link with Arabic language immediately provided children with the key to access to all forms of civil participation, as well as the security of daily stability for the children and their families. Starting from the idea of recreating an environment as close as possible to the country and the culture of origin, the aims of the association are mainly to create a bilingual community, by preserving the Arabic language, including the Turkish one into the educational background, and giving psychological support for integration of Syrian people into social life in Turkey. The fieldwork was characterised by constant adaptations to the many changes in the refugee situation. The integration plan put in place by Turkish authorities presents structural difficulties, which emerge, for instance, in school curricula. They are characterised by a strong sense of nationalism and a general underestimation of linguistic, cultural and identity diversity. The denial of linguistic and cultural identity is a source of conflict and claims. In many cases, this denial slows down or prevents the process of Syrian acceptance of integration, which is experienced as a cancellation of identity and language in favour of the dominant local community. At the same time, in case of a return to Syria, exclusive education in Turkish will prevent an entire generation from accessing professions and roles that require the management of Arabic language in all its components (speaking, understanding, reading and writing). In addition to this there is the Turkish government's failure to build a path with Syrian families that would reduce mistrust towards the Turkish community and gradually make Syrian indefinite stay in the country constructive. "Amal for Education" places language education for refugees at the heart of its philosophy, demonstrating that it is the trump

card for the preservation of Syrian linguistic and cultural identity, on the one hand, and the inclusion and integration into the newTurkish identity, on the other hand.