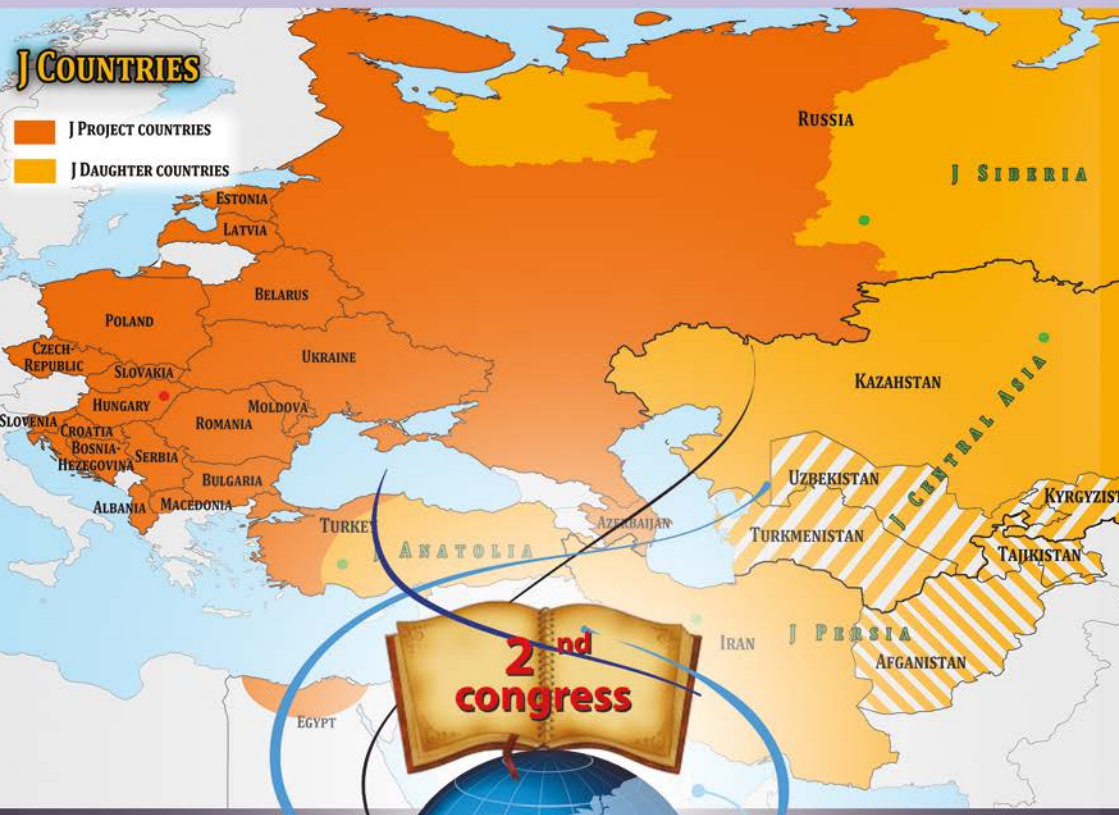

2nd J Project Congress
2-5 March, 2016 I Antalya, Turkey

PROGRAM

PROGRAM

Wednesday, 2 March 2016

10:00-20:00	**Registration**
14:00	**J PROJECT STEERING COMMITTEE MEETING**
17:20-17:30	**OPENING ADDRESSES** *L MARODI, I REISLI*
17:30-19:00	**OPENING LECTURES** **Chairs:** *L MARODI, I REISLI*
17:30	**Novel immunodeficiences causing fungal diseases** *A Puel, Paris, France*
18:15	**Novel defects of T cell differentiation and function** *K Boztug, Vienna, Austria*
19:00	**DINNER**

PROGRAM

Thursday, 3 March 2016

08:30-10:00	**PROGRESS IN UNDERSTANDING B CELL IMMUNODEFICIENCIES I.** **Chairs:** *K WARNATZ, A BERGLOF, S SHARAPOVA*
08:30	**Primary defects of activation and function of B cells** *K Warnatz, Freiburg, Germany*
09:00	**X-linked agammaglobulinemia and phosphoglucomutase 3 deficiency** *A Berglof, Stockholm, Sweden*
09:30	**Activated PIK3DC syndrome: From asymptomatic condition to atypical SCID** *S Sharapova, Minsk, Belarus*
10:00-11:00	**POSTERS 1-15**
11:00-11:30	**BREAK**
11:30-12:30	**PROGRESS IN UNDERSTANDING B CELL IMMUNODEFICIENCIES II.** **Chairs:** *E NAUMOVA, A AGHAMOHAMMADI, A KLOCPERK*
11:30	**Novel genetic findings in patients with CVID in Iran** *A Aghamohammadi, Tehran, Iran*
11:45	**CD27 deficiency** *H Abolhassani, Tehran, Iran*
12:00	**Antibody deficiency in early life and elderly** *E Naumova, Sofia, Bulgaria*
12:15	**Low immunoglobulin levels with no clinical manifestation of PID** *Z Chovancova, Prague, Chech Republic*
12:30-13:00	**POSTERS 16-23**
13:00-14:15	**LUNCH**

14:15-15:45	**DEFECTS OF NEUTROPHILS AND COMPLEMENT I.** **Chairs:** *LT ROUMENINA, N REZAEI, S ROSENZWEIG*
14:15	**Update on primary complement deficiencies** *LT Roumenina, Paris, France*
14:45	**Novel genetic defects of neutrophils and monocytes** *S Rosenzweig, Bethesda, MD*
15:15	**Diagnostic approach to phagocytic cell deficiencies** *N Rezaei, Tehran, Iran*
15:45-16:30	**POSTERS 24-30**
16:30-17:00	**BREAK**
17:00-18:45	**DEFECTS OF NEUTROPHILS AND COMPLEMENT II.** **Chairs:** *A ELMARSAFY, A BOUSFIHA, MY KOKER*
17:00	**Residual activity of NADPH oxidase in patients with CDG patients** *MY Koker, Kayseri, Turkey*
17:15	**Chronic granulomatous disease: The Egyptian experience** *A Elmarsafy, Cairo, Egypt*
17:30	**Renal abscess in HIES 1** *O Paschenko, Moscow, Russia*
17:45	**Congenital neutropenia due to CSF3R mutation** *E Unal, Kayseri, Turkey*
18:00	**IFNγ improves the oxidative burst activity in patients with CGD** *S Filiz, Antalya, Turkey*
18:15	**Unusual clinical manifestation of CGD** *E Heropolitanska-Pliszka, Warsaw, Poland*
18:30	**Novel classification of complement and phagocyte defects** *A Bousfiha, Casablanca, Morocco*
19:00	**DINNER**

PROGRAM

Friday, 4 March 2016

08:30-10:00	**ADVANCES IN UNDERSTANDING T CELL IMMUNODEFICIENCIES I.** **Chairs:** *E BERNATOWSKA, A VILLA, Y SAVCHAK*
08:30	**Clinical manifestations and genetics of Omenn syndrome** *A Villa, Milan, Italy*
09:00	**Tregs and PIDs** *A Sediva, Prague, Czech Republic*
09:20	**Cooperation at J PROJECT levels to increase awareness on NBS** *E Bernatowska, Warsaw, Poland*
09:40	**T Cell PID genetic analysis in Konya** *I Reisli, Konya, Turkey*
10:00-11:15	**POSTERS 31-46**
11:15-11:45	**BREAK**
11:45-13:00	**ADVANCES IN UNDERSTANDING T CELL IMMUNODEFICIENCIES II.** **Chairs:** *A SEDIVA, M BELEVTSEV, Y ROMANYSHYN*
11:45	**Clinical features of patients with *DCLRE1C* mutations** *E Hazar Sayar, Konya, Turkey*
12:00	**IL10R deficiency leading to early onset IBD and cured with HSCT** *NE Karaca, Izmir, Turkey*
12:15	**HSCT in patients with MALT1 deficiency** *G Markelj, Ljubljana, Slovenia*
12:30	**Mucocutaneous aspergillosis associated with DOCK8 deficiency** *Y Romanyshyn, Lviv, Ukraine*
12:45	**EBV-induced lymphoproliferative disease associated with NBS** *Y Savchak, Lviv, Ukraine*

13:00-14:15 **LUNCH**

14:15-15:45 **GENETIC DEFECTS OF CYTOKINE SIGNALING AND IMMUNOREGULATION I.**
Chairs: *ES HUSEBYE, F BENVENUTO, M ALBERT*

14:15 **The extending spectrum of phenotypes in patients with AIRE mutation**
ES Husebye, Oslo, Norway

14:45 **Genetic defects and management of patients with DOCK8 deficiency**
M Albert, Munich, Germany

15:15 **Primary chemokine receptor deficiencies**
F Benvenuto, Trieste, Italy

15:45-16:30 **POSTERS 47-56**

16:30-17:00 **BREAK**

17:00-18:00 **GENETIC DEFECTS OF CYTOKINE SIGNALING AND IMMUNOREGULATION II.**
Chairs: *ES HUSEBYE, F BENVENUTO, B SOLTESZ*

17:00 **Splicing mutations causing XLP**
B Soltesz, Debrecen, Hungary

17:15 **The effect of sirolimus in patients with PI3K pathway defects**
A Kiykim, Istanbul, Turkey

17:30 **SCID presented as HLH**
M Bataneant, Timisoara, Romania

17:45 **J Project - CIS partnership to explore novel PIDs**
L Marodi, Debrecen, Hungary
S Rosenzweig, Bethesda, MD

18:30 **DINNER**

PROGRAM

Saturay, 5 March 2016

09:00-10:55	**INNATE IMMUNE DEFICIENCIES** **Chairs:** *H von BERNUTH, E JOUANGUY, M GURSEL*
09:00	**Innate immune defects and viral infections** *E Jouanguy, Paris, France*
09:30	**Innate immune defects predisposing to bacterial infections** *H von Bernuth, Berlin, Germany*
10:00	**DNA repaır defects and interferonopathies** *M Gursel, Ankara, Turkey*
10:15	**APDS2 syndrome: a long way from clinical presentation to diagnosis** *S Vakhlyarskaya, Moscow, Russia*
10:25	**Di George syndrome: from incidental findings to clinical diagnosis** *I Tuzankina, Ekaterinburg, Russia*
10:35	**Recurrent aseptic abscesses as manifestations in Crohn's disease** *M Fallahpour, Tehran, Iran*
10:45	**Early onset IBD** *M Mesdaghi, Tehran, Iran*
10:55-11:15	**POSTERS 57-64**
11:15-11:45	**BREAK**
11:45-13:15	**GENETICS OF AUTOIMMUNITY AND AUTOINFLAMMATORY SYNDROMES** **Chairs:** *F RIEUX-LAUCAT, M DEBELIAK, B WOLSKA*
11:45	**Inborn errors of autoimmunity** *F Rieux-Laucat, Paris, France*
12:15	**Recent advances in genetics of autoinflammatory syndromes** *M Debeliak, Ljubljana, Slovenia*
12:30	**Pathogen recognition receptor signaling in autoinflammatory disorders** *I Gursel, Ankara, Turkey.*

12:45	**Unclassified systemic autoinflammatory diseases** *B Wolska, Warsaw, Poland*
13:00	**Follow-up on PID patients in a tertiary hospital** *R Molatefi, Tehran, Iran*
13:15-13:55	**POSTERS 65-79**
13:55-14:00	**CLOSING** *L MARODI, I REISLI*
14:00	**LUNCH**

POSTER SESSION I.

B CELLS

1. **Novel BTK mutations in patients with XLA from Eastern and Central Europe**
   B Soltesz, Debrecen, Hungary
2. **Agammaglobulinemia presented with hemiparesis**
   D Hafızoglu, Erzurum, Turkey
3. **Elevated IgE levels in patients with Bruton's disease**
   H Anıl, Eskisehir, Turkey
4. **CVID with pericardial effusion**
   M Khoshkhui, Mashhad, Iran
5. **Long term outcome of patients with CVID**
   S Velbri, Tallinn, Estonia
6. **Occurrence of bronchial asthma in patients with CVID**
   T Milota, Prague, Czech Republic
7. **Case report on childhood CVID**
   A Kiykim, Istanbul, Turkey
8. **Successful lung transplantation in a CVID patient**
   E Gasiuniene, Kaunas, Lithuania
9. **A rare combination of two rare disorders: eosinophilic jejunitis and CVID**
   L Tamasauskiene, Kaunas, Lithuania
10. **Recalcitrant to treatment of IBD in CVID**
    S Soyyigit, Konya, Turkey
11. **Unusual bloodstream infection in a patient with CVID**
    R Ucar, Konya, Turkey
12. **Age-dependency of lymphocyte subpopulations in CVID patients**
    J Litzman, Prague, Czech Republic
13. **HSCT in CVID associated with monosomy 7.**
    S Blazina, Ljubljana, Slovenia
14. **Potentially beneficial effect of hydroxychloroquine in a patient with PIK3CD deficiency**
    A Kiykim, Istanbul, Turkey
15. **Cases of IL-10 receptor deficiency**
    S Burcak Sahin, Konya, Turkey
16. **LRBA gene defect and multiple gallstones**
    S Kutlug, Samsun, Turkey
17. **Selective IgM deficiency**
    Z Chovancova, Prague, Czech Republic
18. **Selective IgM deficiency in an adult with celiac disease and recurrent pneumonia**
    M Heidarzadeh, Kashan, Iran
19. **Clinical characteristics of children with selective IgA deficiency**
    F Orhan, Trabzon, Turkey
20. **Five years of experience in transient hypogammaglobulinemia of infancy**
    N Gulez, Izmir, Turkey
21. **Quality of life of patients with primary antibody deficiencies**
    C Tavakkol, Alborz, Iran
22. **Quality of life in PID patients treated with IVIG or SCIG**
    H Bozkurt, Konya, Turkey
23. **Vaccine-induced pitryasis lichenoides treated with IVIG**
    S Kutlug, Samsun, Turkey

PHAGOCYTES AND COMPLEMENT

24. **CGD patients with brain abscess**
MS Yılmaz, Konya, Turkey
25. **The value of DHR assay in the diagnose CGD**
S Kutlug, Samsun, Turkey
26. **CGD with fungal infections in early infancy**
F Karbasian, Tehran, Iran
27. **Leukocyte adhesion deficiency type 1**
T Prokofjeva, Riga, Latvia
28. **Case series of LAD**
H Gorjipour, Tehran, Iran
29. **DHR assay in MPO deficiency**
M Yavuz Koker, Istanbul, Turkey
30. **Eight years experience with angioedema in north of Iran**
T Cheraghi, Rasht, Iran

T CELLS/SCID

31. **Costimulatory molecule expression in Artemis deficiency**
MA Karaselek, Konya, Turkey
32. **Idiopathic CD4 lymphopenia**
E Hlavackova, Prague, Czech Republic
33. **SCID in a child with adenosine deaminase deficiency**
G. Ristic, Belgrade, Serbia
34. **Case report of a patient with T-B+NK+ deficiency**
N Gesheva, Sofia, Bulgaria
35. **A novel insertion mutation in the IL2RG**
Zs Pistar, Debrecen, Hungary
36. **AR T- B+ NK+ SCID patients: IL-7R is not always the accused gene**
R El-Hawary, Cairo, Egypt
37. **SCID case reports**
P Ciznar, Bratislava, Slovakia
38. **Evaluation of DOCK8 deficient patients: A single centre experience**
A Kiykim, Istanbul, Turkey
39. **Case repor of DOCK8 mutation**
N Moazzan, Mashaad, Iran
40. **HSCT in a child with combined immunodeficiency associated with monosomy 7**
S Blazina, Ljubljana, Slovenia

REGULATORY DEFECTS

41. **Egyptian Children presenting with immunedysregulation**
J Boutros, Cairo, Egypt
42. **Markers in different PID with immunodysregulation**
M Belevtsev, Minsk, Belarus
43. **Hemophagocytic lymphohistiocystosis**
A Volokha, Kiev, Ukraine
44. **Recurrent SH2D1A mutation detected in a 17-year-old male patient**
B Toth, Debrecen, Hungary
45. **Challenges of prenatal genetic diagnosis of FOXP3 mutation**
B Toth, Debrecen, Hungary
46. **FOXP3 expression in primary immunodeficiencies**
S Keyik, Konya, Turkey

PID SYNDROMES

47. **HIES with brain vasculitis**
T Momen, Isfahan, Iran
48. **Case of HIES**
MR Ghaemi Mirabad, Tehran, Iran

49. **Oral and maxillofacial characteristics in sporadic and AD HIES**
M Szegedi, Debrecen, Hungary
50. **Malignant complications in patients with NBS**
I Savchak, Lviv, Ukraine
51. **STAT3 mutation presenting with atopic dermatitis**
S Karaman, Izmir, Turkey
52. **Clinical and laboratory features of Latvian patients with NBS and LAD**
T Prokofjeva, Riga, Latvia
53. **Malignant complications of NBS**
L Kostyuchenko, Lviv, Ukraine
54. **NBS in Eastern Slavs**
M Belevtsev, Minsk, Belarus
55. **SBDS case report**
S Deryabina, Ekaterinburg, Russia
56. **Chromosome 22q11 deletion presenting with immune-mediated cytopenias**
F Genel, İzmir, Turkey

INNATE DEFECTS

57. **Mendelian susceptibility to mycobacterial disease in a center in Iran**
N Nekooie Marnany, Isfahan, Iran
58. **IL-12RB1 mutation in an Egyptian child presents with both: MSMD & ALP**
A Elmarsafy, Cairo, Egypt
59. **Invasive fungal infection due to CARD9 deficiency**
Z Chavoshzadeh, Tehran, Iran
60. **Heterogeneity in presentation of STAT1 mutations**
A Kiykim, Istanbul, Turkey
61. **CMC and STAT1 mutation**
DF Kocacik Uygun, Antalya, Turkey

INFLAMMATORY DISEASES

62. **Patients with inflammatory bowel disease suspected of PID**
B Bagherpour, Isfahan, Iran
63. **Cases of auto-inflammatory diseases in Ukraine**
L. Chernyshova, Kiev, Ukraine
64. **Autoimmune diseases as manifestations of PIDs**
S Blazina, Ljubljana, Slovenia

UNCLASSIFIED

65. **Novel pathogenes isolated from patients with primary immunodeficiencies**
SN Guner, Konya, Turkey
66. **Clinical symptoms of PID with no immunological disorder**
M Skomska, Warsaw, Poland
67. **Pregnancy and childbearing in PID: the matters that need to be looked at**
R Sherkat, Isfahan, Iran
68. **Systemic complications and comorbidities in adult patients with PID**
Z Calıskaner, Konya, Turkey
69. **Epidemiological and clinical features of patients with primary antibody deficiencies**
E Topal, Malatya, Turkey
70. **Genetic causes of bronchiectasis**
SB Erdem, İzmir, Turkey
71. **Rare forms of GVHD and secondary tumor in PID patients**
I Kondratenko, Moscow, Russia
72. **The first Biobank of patients with chronic cutaneous Leishmaniasis suspected of PID**
M Moafi, Isfahan, Iran

73. **Epidemiology of HTLV infections in Iran**
R Farid Hosseini, Mashaad, Iran

REGISTRIES AND NEWBORN SCREENING

74. **National registry of PID in Belarus**
M Belevtsev, Minsk, Belarus
75. **CGD Registry of Turkey**
B Saraymen, Istanbul, Turkey
76. **Phenotypic and genotypic parallels in the regional register of PID**
I Tuzankina, Ekaterinburg, Russia
77. **PID in a highly consanguineous area: It is not always one hit**
S Meshaal, Cairo, Egypt
78. **Selective screening by TREC assay**
V Urdova, Bratislava, Slovakia
79. **Neonatal screening of immunodeficiencies**
MA Bolkov, Ekaterinburg, Russia