▶▶▶ プログラム (Program)

SL　Special Lecture　ENG

Saturday March 23, 2024　15:20-16:10 (Hall)

Chair: Surjit Singh (PGIMER)

SL	**One gene, many phenotypes – RAG deficiency: immune deficiency with immune dysregulation**	Luigi D. Notarangelo(National Institutes of Health)

CL　Chairperson's Lecture　ENG

Saturday March 23, 2024　17:20-18:10 (Hall)

Chair: 森尾 友宏 /Tomohiro Morio (東京医科歯科大学 /Tokyo Medical and Dental University)

CL	**D'où venons-nous? Que sommes-nous? Où allons-nous?**	金兼 弘和(東京医科歯科大学) Hirokazu Kanegane(Tokyo Medical and Dental University)

JSY1　JSIAD シンポジウム 1　拡大新生児スクリーニングを IEI の診療にどう生かすか / JSIAD Symposium 1　Newborn screening for inborn errors of immunity　JPN

Friday March 22, 2024　10:20-12:00 (Hall East)

Chair: 水上 智之 /Tomoyuki Mizukami (国立病院機構熊本医療センター /National Hospital Organization Kumamoto Medical Center)

村松 秀城 /Hideki Muramatsu (名古屋大学医学部附属病院 /Nagoya University Hospital)

JSY1-1	原発性免疫不全症の拡大新生児スクリーニング検査体制	石毛 信之(公益財団法人東京都予防医学協会)
	Expanded newborn screening system for primary immunodeficiency	Nobuyuki Ishige(Tokyo Health Service Association)
JSY1-2	愛知県におけるTRECとKRECによる拡大新生児スクリーニング検査	若松 学(名古屋大学)
	TREC and/or KREC newborn screening programs in Aichi Prefecture	Manabu Wakamatsu(Nagoya University)
JSY1-3	拡大新生児スクリーニングをIEIの診療にどう生かすか　KRECスクリーニング	友政 弾(東京医科歯科大学)
	How to apply expanded newborn screening to IEI treatment: KREC screening	Dan Tomomasa(Tokyo Medical and Dental University)
JSY1-4	**Newborn Screening for Severe Combined Immunodeficiency in Taiwan: A Comprehensive Approach to Early Intervention and Management**	Yin-Hsiu Chien(National Taiwan University Hospital)
JSY1-5	プロテオーム解析による新たな新生児スクリーニング法開発	八角 高裕(京都大学)
	A Non-targeted Proteomics Platform for Newborn Screening of Genetic Disorders	Takahiro Yasumi(Kyoto University)

JSY2　JSIAD シンポジウム 2　成人自己炎症性疾患　最近の話題 / JSIAD Symposium 2　Recent topics in autoinflammatory disorders in adults　JPN

Friday March 22, 2024　13:40-15:10 (Hall East)

Chair: 右田 清志 /Kiyoshi Migita (福島県立医科大学 /Fukushima Medical University)

井田 弘明 /Hiroaki Ida (久留米大学 /Kurume University)

JSY2-1	VEXAS症候群を含めた成人自己炎症性疾患最近の話題	桐野 洋平(横浜市立大学)
	Recent Topics in Adult Autoinflammatory Diseases, Including VEXAS Syndrome	Yohei Kirino(Yokohama City University)
JSY2-2	成人における自己炎症性疾患の診断における留意点 ～バリアントの解釈とその意義について～	古賀 智裕(長崎大学)
	Points to consider in the diagnosis of autoinflammatory diseases in adults - Interpretation of variants and their significance	Tomohiro Koga(Nagasaki University)
JSY2-3	自己炎症性疾患における体細胞モザイクの診断体制の構築：研究から検査へ	小原 收(かずさDNA研究所)
	Establishment of a diagnosis system for somatic mosaicism in autoinflammation: From research to genetic testing	Osamu Ohara(Kazusa DNA Research Institute)
JSY2-4	Clonal hematopoiesisと自己炎症性疾患	井澤 和司(京都大学医学部附属病院)
	Clonal hematopoiesis and autoinflammatory diseases	Kazushi Izawa(Kyoto University Hospital)

ASY1　ASPID Symposium 1　Hematopoietic cell transplantation for inborn errors of immunity　ENG

Sunday March 24, 2024　8:00-9:20 (Hall East)

Chair: Luigi D. Notarangelo (National Institutes of Health)
石村 匡崇 /Masataka Ishimura (九州大学病院 /Kyushu University)

ASY1-1	**HEMATOPOIETIC CELL TRANSPLANTATION FOR INBORN ERRORS OF IMMUNITY: EXPERIENCES IN EUROPE**	Fulvio Porta(Children's Hospital, ASST Spdali Civili, Bresia)
ASY1-2	**Optimizing Outcome of Patients Receiving HSCT for PID – Perspectives in Asia**	Pamela Lee(The University of Hong Kong)
ASY1-3	**Recent advances in allogenic hematopoietic cell transplantation for Japanese patients with inborn errors of immunity**	Satoshi Miyamoto(Tokyo Medical and Dental University (TMDU))

ASY2　ASPID Symposium 2　Basic research of inborn errors of immunity　ENG

Sunday March 24, 2024　10:50-12:10 (Hall West)

Chair: 峯岸 克行 /Yoshiyuki Minegishi (徳島大学 /Tokushima University)
Hans D Ochs (Washington University)

ASY2-1	**CRISPR-Based Treatment of Immunocompromised Host Infections and Generation of Induced Pluripotent Stem-Cell Models of Inborn Errors of Immunity**	David B. Lewis(Stanford University)
ASY2-2	**From Bench to Bedside: Applications of Humanized Mice in the Research of Translational Immunology**	Wenwei Tu(University of Hong Kong)
ASY2-3	**Unwinding the molecular pathogenesis of a novel inherited immunodysregulatory disorder caused by loss-of-function variants in DExD/H box helicase SBNO2**	Kazuyuki Meguro(National Institutes of Health, Chiba University Hospital)

JEP　JSIAD 優秀演題 YIA/JSIAD Excellent Presentation　JPN

Saturday March 23, 2024　13:10-14:10 (Hall East)

Chair: 小野寺 雅史 /Masafumi Onodera (国立成育医療研究センター / National Center for Child Health and Development)
増本 純也 /Junya Masumoto (愛媛大学 /Ehime University)

JEP-1	クリオピリン関連周期熱症候群における体細胞モザイク変異率の推移とシングルセル解析による病態解明	加藤 健太郎(京都大学)
	Longitudinal trends of somatic mutation rates and single-cell functional analysis in cryopyrin-associated periodic syndrome somatic mosaic patients	Kentaro Kato(Kyoto University)
JEP-2	中條-西村症候群のモデルとしての*Psmb8* ^{G201V}変異ノックインマウスの作製と特性評価	原 知之(和歌山県立医科大学)
	Generation and characterization of the *Psmb8 G201V* mutation knock-in mice as a model for Nakajo-Nishimura syndrome	Tomoyuki Hara(Wakayama Medical University)
JEP-3	ESCRT複合体の機能不全はcGAS/STING経路に依存した炎症応答を引き起こす	砂山 風磨(東北大学)
	Dysfunction of the ESCRT complex leads to cGAS/STING-dependent inflammatory responses	Fuma Sunayama(Tohoku University)
JEP-4	**Induction of Eomes-expressing Th cells via upregulation of type I interferon**	Tzuwen Yeh(National Center of Neurology and Psychiatry)

AEP　APSID YIA Presentation　ENG

Sunday March 24, 2024　9:30-10:30 (Hall East)

Chair: Yu-Lung Lau (Hong Kong University)
Narissara Suratannon (Chulalongkorn University)

AYIA-1	**Outcome of HSCT in children with rare inborn errors of immunity-Ten years experience from a tertiary referral centre**	Venkateswaran Vellaichamy Swaminathan(Apollo Hospitals)
AYIA-2	**Tregopathies: Single centre experience of 25 patients with monogenic autoimmunity**	Vaishnavi Venkatachari Iyengar(BJ wadia hospital for children)
AYIA-3	**Detection of copy number variations in Primary immunodeficiency Disease using Multiplex Dependent Probe Amplification**	Anit Kaur(Advanced Pediatric Centre)
AYIA-4	**Exploring Inborn Errors of Immunity with Neutropenia: Clinical and Molecular Insights from North India**	Rakesh Kumar Pilania(PGIMER)

IPOPI　IPOPI Joint Session　PID Life Odyssey – the Patient Journey　ENG

Sunday March 24, 2024　15:30-16:30 (Hall West)

Moderator: Nizar Mahlaoui (l'hôpital universitaire Necker-Enfants Malades)
Martin van Hagen (Erasmus University Medical Center)

JEL1　教育セッション 1/Educational Lecture 1　JPN

Friday March 22, 2024　9:10-9:40 (Hall West)

Chair: 小野寺 雅史 /Masafumi Onodera (国立成育医療研究センター / National Center for Child Health and Development)

JEL1	PIDJ2の概要と今後の展望～データ利活用に向けて～	今井 耕輔(防衛医科大学校)
	PIDJ2: Primary Immunodeficiency Database in Japan ver.2 ~ Overviewand future perspectives towards data utilization	Kohsuke Imai(National Defense Medical College)

JEL2　教育セッション 2/Educational Lecture 2　JPN

Friday March 22, 2024　16:30-17:30 (Hall West)

Chair: 高田 英俊 /Hidetoshi Takada (筑波大学 /Tsukuba University)

JEL2-1　フローサイトメーター検査結果の解釈のしかた　星野 顕宏(東京医科歯科大学)

A practical approach to flow cytometric analysis　Akihiro Hoshino(Tokyo Medical and Dental University)

JEL2-2　PIDJ の仕組みと遺伝子検査の出し方　岡田 賢(広島大学)

The system of PIDJ and the practical implementation of genetic test in IEI　Satoshi Okada(Hiroshima University)

JOP1　JSIAD 口演 1　疫学・全国調査 / JSIAD Oral Presentation 1　Epidemiology/National Survey　JPN

Friday March 22, 2024　9:10-10:10 (Hall East)

Chair: 藤尾 圭志 /Keishi Fujio (東京大学 /Tokyo University)

森 雅亮 /Masaaki Mori (聖マリアンナ医科大学 /St. Marianna University School of Medicine)

JOP1-1　自己炎症性疾患の移行期医療の現状：会員アンケート結果から　日高 由紀子(久留米大学)

Current Status of Transitional Care for Autoinflammatory Diseases: Results of a Member Questionnaire　Yukiko Hidaka(Kurume University)

JOP1-2　日本の原発性免疫不全症候群患者を対象としたPatient Reported Outcomes研究―非介入前向き観察研究―中間解析報告　河合 利尚(国立成育医療研究センター)

Patient-Reported Outcomes in Patients with Primary Immunodeficiency Diseases in Japan: Baseline Results from a Prospective Observational Study　Toshinao Kawai(National Center for Child Health and Development)

JOP1-3　原発性免疫不全症の移行支援の現状調査　河合 利尚(国立成育医療研究センター)

Transition to Adult Care in Inborn Errors of Immunity: a web-based study　Toshinao Kawai(National Center for Child Health and Development)

JOP1-4　健常妊産婦における血漿補体値の推移についての検討　辻本 弘(和歌山県立医科大学)

Measurement of plasma complement levels in pregnant women　Hiroshi Tsujimoto(Wakayama Medical University)

JOP1-5　X連鎖無ガンマグロブリン血症の当院における後方視的検討 -新生児スクリーニングの重要性に関して-　井上 翔太(防衛医科大学校病院)

Importance of Newborn Screening Revealed by a Retrospective Study of X-linked agammaglobulinemia in a single hospital　Shota Inoue(National Defense Medical College)

JOP1-6　茨城県における新生児TREC/KRECスクリーニング　穂坂 翔(筑波大学附属病院)

TREC/KREC Newborn Screening in Ibaraki Prefecture　Sho Hosaka(University of Tsukuba Hospital)

JOP2　JSIAD 口演 2　抗体産生不全症 / JSIAD Oral Presentation 2　Primary antibody deficiency　JPN

Friday March 22, 2024　9:50-10:50 (Hall West)

Chair: 竹崎 俊一郎 /Shunichiro Takesaki (北海道大学 /Hokkaido University)

松田 裕介 /Yusuke Matsuda (金沢大学 /Kanazawa University)

JOP2-1　*NFKB1*遺伝子異常を伴う分類不能型免疫不全症(common variable immunodeficiency: CVID)の1例　三宅 淳(久留米大学)

A case of common variable immunodeficiency (CVID) with *NFKB1* gene abnormality　Atsushi Miyake(Kurume University)

JOP2-2	青年期まで無症状で経過した*NFKB2*ハプロ不全によるDAVID症候群の1例	内田 嶺花(東京医科歯科大学)
	A case of asymptomatic DAVID syndrome with haploinsufficiency of *NFkB2* mutation diagnosed in adolescent	Reika Uchida(Tokyo Medical and Dental University)
JOP2-3	中耳炎を繰り返し、低ガンマグロブリン血症とT細胞機能低下を契機に診断されたXMEN病	白潟 爽香(国立成育医療研究センター)
	XMEN disease diagnosed after recurrent otitis media, hypogammaglobulinemia and decreased T-cell function	Sayaka Shiragata(National Center for Child Health and Development)
JOP2-4	分類不能型免疫不全症および小児急性B細胞性リンパ性白血病を発症した患者における*PAX5*の生殖系列ヘテロバリアント	宮本 智史(東京医科歯科大学)
	A *PAX5* heterozygous germline variant in a patient with common variable immunodeficiency complicated with pediatric B-cell acute lymphoblastic leukemia	Satoshi Miyamoto(Tokyo Medical and Dental University)
JOP2-5	*TCF3* E555K変異の分子病態と特殊性に対する検証	内海 孝法(広島大学)
	Validation of *TCF3* E555K Variant against Molecular Mechanisms and Unique Characteristics	Takanori Utsumi(Hiroshima University)
JOP2-6	Late onset combined deficiency(LOCID)を呈した18q欠失症候群	橋口 祥(やまびこ医療福祉センター)
	18q deletion syndrome presenting with late onset combined immunodeficiency	Sho Hashiguchi(Yamabiko Medical Welfare Center)

JOP3　JSIAD 口演 3　高 IgE 症候群・慢性肉芽腫症 / JSIAD Oral Presentation 3　Hyper-IgE syndrome/chronic granulomatous disease　JPN

Friday March 22, 2024　11:00-12:00 (Hall West)

Chair: 河合 利尚 /Toshinao Kawai (国立成育医療研究センター / National Center for Child Health and Development)
山田 雅文 /Masafumi Yamada (酪農学園大学 /Rakuno Gakuen University)

JOP3-1	*STAT3*遺伝子に新規バリアントが検出された常染色体顕性遺伝高IgE症候群の親子例	東川 朋子(松阪中央総合病院)
	A novel hypomorphic variant in the *STAT3* gene identified in a 7-year-old male with hyper-IgE syndrome	Tomoko Higashigawa(Matsusaka Chuo General Hospital)
JOP3-2	頚部化膿性リンパ節炎を繰り返したため皮下注用ヒト免疫グロブリン製剤を導入した高IgE症候群(STAT3-LoF)の一例	西口 雅人(岐阜大学)
	Subcutaneous immunoglobulin replacement therapy was effective against recurrent cold suppurative granulomatous lymphadenitis with hyper-IgE syndrome: a case report	Masato Nishiguchi(Gifu Unversity)
JOP3-3	肝内仮性動脈瘤による胆道出血を生じ、その後の全身スクリーニング検査で無症候性右冠動脈瘤を認めたSTAT3-高IgE症候群の一例	藤田 大輝(帯広厚生病院)
	Biliary hemorrhage by intrahepatic pseudoaneurysm and asymptomatic right coronary aneurysm in a patient with STAT3 Hyper IgE Syndrome	Daiki Fujita(Obihiro-Kosei General Hospital)
JOP3-4	$p67^{phox}$欠損慢性肉芽腫症に1型糖尿病を合併した14歳女児例	芹澤 陽菜(北里大学病院)
	A case of 14 years old girl who was diagnosed with type1 diabetes and $p67^{phox}$ deficient chronic granulomatous disease	Haruna Serizawa(Kitasato University School of Medicine)
JOP3-5	当科における慢性肉芽腫症腸炎の臨床像と内視鏡所見	松田 裕介(金沢大学)
	Clinical presentation and endoscopic findings of CGD colitis : a single center experience	Yusuke Matsuda(Kanazawa University)
JOP3-6	先天性好中球減少症と骨髄線維症から重症先天性好中球減少症5型と診断した1歳女児の診療経過と病態解析	髙橋 和樹(北海道大学病院)
	Clinical course and immunological analysis of a girl with congenital neutropenia and myelofibrosis diagnosed with severe congenital neutropenia type 5	Kazuki Takahashi(Hokkaido University Hospital)

JOP4　JSIAD 口演 4　自己抗体産生異常症その他 /
JSIAD Oral Presentation 4　Diseases of autoimmunity and immune dysregulation　JPN

Friday March 22, 2024　13:40-14:40 (Hall West)

Chair: 宮前 多佳子 /Takako Miyamae (東京女子医科大学 /Tokyo Women's Medical University)
植木 将弘 /Masahiro Ueki (北海道大学病院 /Hokkaido University Hospital)

JOP4-1	成人期から抗TNFα抗体を開始したADA2欠損症の家族例	穂坂 翔(筑波大学附属病院)
	Two related cases of ADA2 deficiency who began anti TNF-α therapy in adulthood	Sho Hosaka(University of Tsukuba Hospital)
JOP4-2	ELF4欠損症患者家系についての報告	荻野 諒(京都大学医学部附属病院)
	Report on a family with DEX (defciency in ELF4, X-linked)	Ryo Ogino(Kyoto University Hospital)
JOP4-3	EBV関連疾患における抗IL27自己抗体の検出	谷田 けい(イマジン研究所)
	Detection of anti-IL27 autoantibodies in EBV-related diseases	Kay Tanita(Institut Imagine)
JOP4-4	STIM依存的カルシウム流入が胚中心B細胞の正の選択と親和性成熟を制御する	矢田 裕太郎(九州大学)
	STIM-mediated store-operated calcium entry regulates positive selection and affinity maturation of germinal center B cells	Yutaro Yada(Kyushu University)
JOP4-5	*CARD11*遺伝子変異によるB細胞増多を認めた1例	西之園 翼(名古屋大学)
	A case of B-cell proliferation due to *CARD11* mutations	Tsubasa Nishinosono(Nagoya University)
JOP4-6	SARS-CoV-2感染を契機に非典型溶血性尿毒症症候群を発症したC3異常症の1例	安藤 正人(岐阜大学)
	Atypical hemolytic uremic syndrome with C3 variant following SARS-CoV-2 infection: A case report	Masato Ando(Gifu University)

JOP5　JSIAD 口演 5　自然免疫異常症その他 /
JSIAD Oral Presentation 5　Defects in innate immunity　JPN

Friday March 22, 2024　15:20-16:20 (Hall East)

Chair: 浅野 孝基 /Takaki Asano (広島大学 /Hiroshima University)
高田 紗奈美 /Sanami Takada (東京医科歯科大学 /Tokyo Medical and Dental University)

JOP5-1	腫瘤形成性多発骨病変で発症し全身型の若年性黄色肉芽種との鑑別を要したBCG骨髄炎を合併した*STAT1*異常症	小寺 麻実(東北大学病院)
	***STAT1* deficiency complicated with BCG osteomyelitis mimicking Systemic Juvenile Xanthogranuloma and developing mass-forming multiple bone lesions**	Asami Kodera(Tohoku University Hospital)
JOP5-2	網羅的機能評価に基づく*RELA*ナンセンス変異の病的意義の法則性の発見	早川 博子(広島大学)
	Comprehensive functional study based discovery of the regularities in the pathological significance of *RELA* nonsense mutations	Hiroko Hayakawa(Hiroshima University)
JOP5-3	外科治療が有効であった超早期発症型炎症性腸疾患の小児例	畠野 真帆(東京医科歯科大学病院)
	A pediatric case of very early-onset inflammatory bowel disease successfully treated with a surgical intervention	Maho Hatano(Tokyo Medical and Dental University Hospital)
JOP5-4	大腸の広範囲にリンパ濾胞過形成を認めたA20ハプロ不全症	宇佐美 雅章(金沢大学)
	A case of a 20 haploinsufficiency with extensive lymphofollicular hyperplasia of the colon	Masaaki Usami(Kanazawa University)
JOP5-5	父親の病歴が手がかりとなり診断に至ったPAMI症候群の一例	住吉 孝允(岐阜大学)
	A case of PAMI syndrome the father's medical history led to the diagnosis	Takamasa Sumiyoshi(Gifu University)

JOP5-6 白血球・好中球数減少と細菌感染症の反復を契機に診断し、TREC・KREC低値であったWHIM症候群の乳児例
東 悠太(函館中央病院)

Low TREC and KREC levels in the patient with WHIM syndrome
Yuta Azuma(Hakodate Central Hospital)

JOP6 JSIAD 口演 6 自己炎症性疾患 / JSIAD Oral Presentation 6 Autoinflammatory disorders

JPN

Friday March 22, 2024 15:10-16:20 (Hall West)

Chair: 日高 由紀子 /Yukiko Hidaka (久留米大学 /Kurume University)
保田 晋助 /Shinsuke Yasuda (東京医科歯科大学 /Tokyo Medical and Dental University)

JOP6-1 自己炎症性乳児期発症腸炎(AIFEC) で発症したマクロファージ活性化症候群(MAS) 関連バリアントをもつNLRC4異常症の一例
加藤 大吾(千葉県こども病院)

A case of NLRC4 inflammasomopathies with macrophage activation syndrome (MAS)-associated variant in autoinflammation with infantile enterocolitis (AIFEC)
Daigo Kato(Chiba Children's Hospital)

JOP6-2 自己炎症性角化症が疑われる皮膚変性病変を認めた13歳女子例
谷 諭美(東京女子医科大学病院)

A 13-year-old girl with degenerative skin lesions suspected to be autoinflammatory keratinization disease
Yumi Tani(Tokyo Women's Medical University Hospital)

JOP6-3 Interferonopathyを来す自己炎症疾患との鑑別に苦慮した抗NXP2抗体陽性若年性特発性炎症性筋疾患の小児例
川邊 智宏(東京女子医科大学)

A child with anti-NXP2 antibody-positive juvenile idiopathic inflammatory myopathy arduous to distinguish from autoinflammatory interferonopathy
Tomohiro Kawabe(Tokyo Women's Medical University)

JOP6-4 Sideroblastic anemia with Immunodeficiency, fevers and developmental delay (SIFD)の一例における小胞体ストレスの解析
長森 恒久(旭川医科大学)

A case of Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay and the evaluation of endoplasmic reticulum stress
Tsunehisa Nagamori(Asahikawa Medical University)

JOP6-5 自然免疫分子STINGのリソソームミクロオートファジー分解を制御する分子の同定
佐藤 加奈子(東北大学)

Identification of lysosome-associated proteins that regulate the microautophagic degradation of STING
Kanako Sato(Tohoku University)

JOP6-6 自然免疫分子STING主要バリアントの機能評価
小出 頌悟(東北大学)

Activites of primary variants of innate immune protein STING
Shogo Koide(Tohoku University)

JOP6-7 VEXAS症候群の患者末梢血を用いたDAMPsと細胞死の機能解析
安達 聡一郎(横浜市立大学)

Functional analysis of DAMPs and cell death using peripheral blood of patients with VEXAS syndrome
Soichiro Adachi(Yokohama City University)

JOP7 JSIAD 口演 7 造血細胞移植 / JSIAD Oral Presentation 7 Hematopoietic cell transplantation

JPN

Friday March 22, 2024 16:30-17:30 (Hall East)

Chair: 高木 正稔 /Masatoshi Takagi (東京医科歯科大学 /Tokyo Medical and Dental University)
森谷 邦彦 /Kunihiko Moriya (防衛医科大学校 /National Defense Medical College)

JOP7-1 新生児スクリーニング検査で同定されたRORγT欠損症
西村 豊樹(宮崎大学)

RORγT deficiency identified by newborn screening
Toyoki Nishimura(Miyazaki University)

JOP7-2 Mcleod症候群を併発した慢性肉芽腫症に対する造血幹細胞移植
藤森 倢太郎(国立成育医療研究センター)

Successful transplantation in a patient chronic granulomatous disease and McLeod phenotype
Kentaro Fujimori(National Center of Child Health and Development)

JOP7-3 ニューモシスチス肺炎, 難治性下痢症を呈したMHCクラスⅡ欠損症の乳児例 — 神山 裕二(横浜市立大学附属病院)

Case Report: An infant of major histocompatibility complex(MHC) classⅡdeficiency with Pneumocystis pneumonia and intractable diarrhea — Yuji Kamiyama(Yokohama City University Hospital)

JOP7-4 鹿児島県拡大新生児マススクリーニング検査を契機に診断されたX-連鎖重症複合免疫不全症 — 西川 拓朗(鹿児島大学)

X-linked severe combined immunodeficiency syndrome identified by extended newborn screening in Kagoshima prefecture — Takuro Nishikawa(Kagoshima University)

JOP7-5 新生児マススクリーニングでTRECの低下を認め, *FOXN1*遺伝子にヘテロ接合性に新規バリアントを認めた双胎例 — 松田 諒(佐世保市総合医療センター)

Twins with heterozygous novel mutation in *FOXN1* gene identified by low copies of TREC in newborn screening — Ryo Matsuda(Sasebo City General Hospital)

JOP7-6 XIAP欠損症におけるアレムツズマブを用いた同種造血細胞移植の後方視解析 — 宮本 智史(東京医科歯科大学)

Allogenic HCT with alemtuzumab for patients with XIAP deficiency — Satoshi Miyamoto(Tokyo Medical and Dental University)

AOP1 APSID Oral Presentation 1 Autoinflammatory 1 ENG

Saturday March 23, 2024 8:30-9:50 (Hall East)

Chair: Huawei Mao (Beijing Children's Hospital)
井田 弘明 /Hiroaki Ida (久留米大学 /Kurume University)

AOP1-1 **TNF Receptor Associated Protein 1 variant predisposing to opportunistic *Pneumocystis jiroveci* infection and respiratory failure in non-HIV subject** — Louis Chai(National University Hospital)

AOP1-2 **Vitronectin a novel urinary proteomic biomarker promotes cell pyroptosis in juvenile systemic lupus erythematosus** — Zhe Cai(Guangzhou Women and Children's Medical Center)

AOP1-3 **NLRP12-associated systemic autoinflammatory disease presented with atypical Kawasaki disease: a case report** — Pin-Chia Huang(National Taiwan University Hospital)

AOP1-4 **ROSAH (retinal dystrophy, optic nerve edema, splenomegaly, anhidrosis, and headache) syndrome with Alpha-protein kinase 1** — Areum Shin(Sungkyunkwan University)

AOP1-6 **A Not-so-severe Congenital Neutropenia** — Donald Vinh(McGill University Health Centre)

AOP1-7 **Atypical familial hemophagocytic lymphohistiocytosis 3 patients** — Xi Yang(Children's Hospital of Chongqing Medical University)

AOP1-8 **RAG1 mutation: from SCID to autoinflammatory manifestations** — Anh Thi Van Nguyen(Vietnam National Children's Hospital)

AOP2 APSID Oral Presentation 2 B-cell deficiency ENG

Saturday March 23, 2024 8:30-9:50 (Hall West)

Chair: Hans D Ochs (Washington University)
遠藤 明史 /Akifumi Endou (東京医科歯科大学 /Tokyo Medical and Dental University)

AOP2-1 **Outcomes of X-linked agammaglobulinaemia patients in the United Kingdom and Hong Kong** — Jaime S Rosa Duque(The University of Hong Kong)

AOP2-2 **BACH-2 Deficiency: Clinical and Immunological Features in a Patient early -onset with Recurrent Infections and Normal Laboratory Test Results** — Mohammadreza Shafiei(Alborz University of Medical Sciences)

AOP2-3	**Impact of Novel *SLC5A6* Mutations on Vaccine Response and B Cell Repertoire Dynamics via GC-Dependent and -Independent Pathways**	Chi-Chang Shieh(National Cheng Kung University Medical College)
AOP2-4	**Autosomal recessive agammaglobulinemia caused by a novel homozygous CD79a mutation due to segmental uniparental disomy of chromosome 19**	Lang Yu(Chongqing Medical University)
AOP2-5	**Agammaglobulinemia, absent tonsils, bronchiectasis, poor humoral immune responses and abnormal naïve helper T-cells associated with a De Novo IKZF1 mutation**	Intan Hakimah Ismail(Universiti Putra Malaysia)
AOP2-6	**Common variable immunodeficiency (CVID) complicated by Crohn's disease in a 6-year-old boy**	Takashi Fujiwara (National Defense Medical College)
AOP2-7	**Novel variants in *SLC39A7* in girl manifested with fever and multiple ecthyma gangrenosum**	Suravat Homvises(King Chulalongkorn Memorial Hospital)
AOP2-8	**X-Linked *SASH3* deficiency presenting with severe varicella infections, recurrent sinopulmonary tract infections and low IgM levels**	Tanatchabhorn Soponkanabhorn (King Chulalongkorn Memorial Hospital)

AOP3 APSID Oral Presentation 3 Autoinflammatory 2 ENG

Saturday March 23, 2024 13:10-14:10 (Hall West)

Chair: Vignesh Pandiarajan (PGIMER)

保田 晋助 /Shinsuke Yasuda (東京医科歯科大学 /Tokyo Medical and Dental University)

AOP3-1	**The genetic and clinical characteristics and effects of Canakinumab on cryopyrin-associated periodic syndrome: a large pediatric cohort study from China**	Zhou Shu(Beijing Children's hospital)
AOP3-2	**Heterozygous NRLP12 Genetic Variant Associated with Chronic Granulomatous Disease-Phenotype**	Ho Wai Koo(Hospital Sultan Abdul Halim)
AOP3-4	**Three *NBAS* deficiency cases highlighting the impact of variant location on clinical outcomes**	Yada Sirisa(King Chulalongkorn Memorial Hospital)
AOP3-5	**Autoinflammatory Syndromes at PGIMER, Chandigarh, India: A Decade of Clinical Insights and Challenges**	Deepti Suri(PGIMER)
AOP3-6	**Activation of NLRP1 Inflammasome Due to Homozygous Deletion of CARD8 in a Patient**	Rui Gan(Children's Hospital of Chongqing Medical University)

AOP4 APSID Oral Presentation 4 MSMD ENG

Sunday March 24, 2024 8:00-9:00 (Hall West)

Chair: Yunfei An (Children's Hospital of Chongqing Medical University)

岡田 賢 /Satoshi Okada (Hiroshima University)

AOP4-1	**IMMUNODEFICIENCY IN AN ADULT PRESENTING AS RECURRENT MYCOBACTERIAL TUBERCULOSIS INFECTION**	Karol Anne Basallo Camonayan-Flor (Philippine Society of Allergy)
AOP4-2	**Autoantibodies to interferon-gamma: clinical features, solutions to diagnostic challenges, and a proposal for in-vitro monitoring of disease**	Valerie Chiang(Queen Mary Hospital)
AOP4-3	**Spectrum of Phagocytic and other Innate Immune Defects from a single center in India**	Akshaya Sanjay Chougule(B. J. Wadia Hospital for Children)
AOP4-4	**Anti-IFN-w autoantibodies and severe adenovirus infection**	Chen-Yen Kuo(Chang Gung University)

AOP4-5	**Interferon alpha therapy in children with Mendelian susceptibility to mycobacterial disease: A case series**	Vaishnavi Venkatachari Iyengar(BJ Wadia Hospital for Children)
AOP4-6	**A full STAT1-mutated lentiviral library constructed by Crispr-Cas9 conducts promising diagnostic value on the classification of STAT1-mutated diseases**	Huilin Mu(Children's hospital of Chongqing Medical University)

AOP5 APSID Oral Presentation 5 IEI susceptible to EBV

ENG

Sunday March 24, 2024 9:30-10:30 (Hall West)

Chair: David Lewis (Stanford Univeristy)

笹原 洋二 /Yoji Sasahara (東北大学 /Tohoku University)

AOP5-1	**New Presentation of CD27 deficiency; Coronary Ectasia and COVID-19**	Samin Sharafian(Mofid Children's Hospital)
AOP5-2	**RAS GUANYL-RELEASING PROTEIN 1(RASGRP1) MUTATION ASSOCIATED WITH DIFFUSE MESANGIAL SCLEROSIS INFANTILE NEPHROTIC SYNDROME AND EPSTEIN-BARR VIRUS (EBV)-INDUCED HODGKIN'S LYMPHOMA**	Khairoon Nisa Mohamed Nashrudin(Universiti Putra Malaysia)
AOP5-3	**A case of *NRAS* mutation associated early onset SLE with multiple infections**	Qi Zheng(Zhejiang University Children's Hospital)
AOP5-4	**Immunodeficiency-associated vaccine-derived poliovirus type 2 (iVDPV2) in a patient with combined primary immunodeficiency**	Joan Camille Calma Sta. Ana(Philippine General Hospital)
AOP5-5	**Spectrum of EBV cohort from Western India- a single centre experience**	Vijaya Gowri(B.J. Wadia Hospital for Children)
AOP5-6	**NOVEL IMMUNODEFICIENCY CAUSED BY HOMOZYGOUS MUTATION IN SOLUTE CARRIER FAMILY 19 MEMBER 1 ENCODING THE REDUCED FOLATE CARRIER**	Akira Shiraishi(Kyushu University)

AOP6 APSID Oral Presentation 6 Basic

ENG

Sunday March 24, 2024 11:10-12:10 (Hall East)

Chair: Elena WY Hsieh(Colorado University)

増本 純也 /Junya Masumoto (愛媛大学 /Ehime University)

AOP6-1	**Effects of WASp on the inhibitory function of Treg by regulating CTLA-4 mediated transendocytosis**	Wenjing Zhang(Children's Hospital of Chongqing Medical University)
AOP6-2	**Global transcriptome profiling of activated T-cells in X-HIGM patients using RNA-sequencing approach**	Manpreet Dhaliwal(Advanced Paediatrics Centre)
AOP6-3	**Autosomal Dominant IRF3 Deficiency in a child with Life-Threatening Enterovirus 71 Encephalitis**	Kang Chen Xuan(Chang Gung University)
AOP6-6	**Patient-derived EPSC platform in STAT1-GoF modelling**	Jane Chi Yan Wong(University of Hong Kong)
AOP6-7	**Palmitoylation restricts SQSTM1/p62-mediated autophagic degradation of NOD2 to modulate inflammation**	Zeng Huasong(Guangzhou Women and Chidren's Medical Center)
AOP6-8	**Novel STAT1 deletion mutations lead to GOF phenotype in two patients displaying atypical clinical spectrum concerning autoimmunity**	Ran Chen(Children's hospital of Chongqing Medical University)

AOP7 APSID Oral Presentation 7 HCT for IEI

ENG

Sunday March 24, 2024 13:45-14:55 (Hall East)

Chair: Fulvio Porta (Children's Hospital Brescia)
森谷 邦彦 /Kunihiko Moriya (防衛医科大学校 /National Defense Medical College)

AOP7-2	**EXCELLENT OUTCOMES IN HSCT FOR CHILDREN WITH INBORN ERROR OF IMMUNITY – EXPERIENCE FROM TERTIARY REFERRAL CENTRE IN INDIA**	Venkateswaran Vellaichamy Swaminathan(Apollo Hospitals)
AOP7-3	**Pretransplant ribavirin and interferon-α therapy for rhinovirus interstitial pneumonia in a RAG1-deficient infant; against respiratory viral infection of HCT**	Nobutaka Harada(Kyushu University)
AOP7-4	**MHC-classII deficiency among Iranian patients : confirmation of c.162delG RFXANK Founder Mutation in the Iranian Population**	Mohadese Sadat Mousavi Khorshidi(Tehran University)
AOP7-6	**Results of haploidentical transplant in patients with donor specific antibodies: a case report**	Le Nguyen-Ngoc-Quynh(Vietnam National Children's Hospital)
AOP7-7	**Effect of Upadacitinib for Clinical Remission in Patient with Crohn's Disease : a Systematic Review of Randomized Controlled Trials**	Mohammad Zuhriansyah Sabran (Pelita Harapan University)

AOP8 APSID Oral Presentation 8 Patient care

ENG

Sunday March 24, 2024 13:45-15:05 (Hall West)

Chair: Yae-Jean Kim (Samsung Medical Center)
大西 秀典 /Hidenori Ohnishi (岐阜大学 /Gifu University)

AOP8-1	**Prevalence And Varieties of Primary Immunodeficiency Disorders among Persistent Pneumonia Cases in Children- A Hospital- Based Study**	Rumana Parveen(Bangladesh Shishu Hospital & Institute)
AOP8-2	**Revolutionizing Lymphocyte Subset Quantification with LySIM: A Cost-Effective and Broadly Accessible Method**	Henkie Isahwan Ahmad Mulyadi Lai(University College of MAIWP International)
AOP8-3	**Cost-Utility analysis of newborn screening for Severe Combined Immunodeficiency (SCID) in Korea**	Jong Youn Moon(Gachon University)
AOP8-4	**Clinical profile and management of pediatric hereditary angioedema in resource constrained settings:our experience from a single centre in North India**	Prabal Barman(PGIMER)
AOP8-5	**Quantifying and visualising our understanding of C1 inhibitor deficiency: A 50-year bibliometric analysis on global research productivity, collaboration and focus**	Hugo W.F. Mak(Queen Mary Hospital)
AOP8-6	**Investigating the performance of ten warning signs of Primary Immunodeficiency as early diagnostic tool: a Nationwide Survey in Japan**	Takahiro Kido(University of Tsukuba Hospital)
AOP8-7	**Profile of Autoimmunity and Immune Dysregulation in 360 Patients with Inborn Errors of Immunity at A Tertiary Care Center In South India**	Neha Singh(Aster CMI Hospital)
AOP8-8	**Characteristics and outcomes of common variable immunodeficiency patients in a tertiary adult institution**	Xin Rong Lim(Tan Tock Seng Hospital)

APP1　APSID Poster-1

ENG

Saturday March 23, 2024　14:20-15:20 (Terrace Room)

Chair: Intan Hakimah Ismail (University Putra Malaysia)
神戸 直智 /Naotomo Kambe (京都大学 /Kyoto University)

APP1-1	**Allo-immune Hemolytic Disease of the Newborn resulting from Rhesus Incompatibility -A Case Report**	Lytheang Try(National Pediatric Hospital)
APP1-2	**Efficacy of JAK inhibitors in Patients with STING-Associated Vasculopathy with Onset in Infancy**	Mengyue Deng(Beijing Children's Hospital)
APP1-3	**Tocilizumab Effectively Managed for a Girl with A20 Haploinsufficiency**	Dae Chul Jeong(The Catholic University of Korea)
APP1-4	**Identified mutation in *FOXP3* gene in Vietnamese patient with IPEX syndrome**	Mai Thi Phuong Nguyen(Vietnam National Children's Hospital)
APP1-5	**Wiskott–Aldrich syndrome protein maintains regulatory T cell tolerance by modulating their surface IL-2 receptor levels**	Zhou Shu(Beijing Children's Hospital)
APP1-6	**Short stature, hypothyroidism and systemic lupus erythematosus: clinical clues to an underlying type 1 interferonopathy**	Prabal Barman(PGIMER)
APP1-7	**Elucidating the Impact of STAT1 Gain-of-Function Mutations: Immunophenotyping and Therapeutic Insights from Chronic Mucocutaneous Candidiasis Cases in Taiwanese Families**	Wei Te Lei(Hsinchu Municipal Mackay Children's Hospital)

APP2　APSID Poster-2

ENG

Saturday March 23, 2024　14:20-15:20 (Terrace Room)

Chair: Xi Yang (Children's Hospital of Chongqing Medical University)
植木 将弘 /Masahiro Ueki (北海道大学病院 /Hokkaido University Hospital)

APP2-1	**Increasing trend of COVID-19 anti-S-RBD and surrogate neutralizing antibodies in X-linked agammaglobulinaemia patients receiving Ig replacement from 2021 to 2023**	Jaime S Rosa Duque(The University of Hong Kong)
APP2-2	**Mild clinical phenotype of Omicron infection in children with inborn errors of immunity**	HanYang(Capital Medical University)
APP2-3	**Identification mutations of *BTK* Gene in patients with X-Linked Agammaglobulinemia Disease**	Tien Manh Ngo(Vietnam National Children's Hospital)
APP2-4	**Predictors of infections in patients with secondary hypogammaglobulinemia in a tertiary rheumatological unit**	Justina Wei Lynn Tan(Tan Tock Seng Hospital Singapore)
APP2-5	**Unmasking the Impact of STAT-1 Gain-of-Function Mutations**	Brenda Guendulain velazquez(Mexican Social Security Institute)
APP2-6	**The first case of chronic osteomyelitis by fluconazole-resistant Candida albicans in a CARD-9 deficiency Korean patient**	Yoonsun Yoon(Korea University Guro Hospital)
APP2-7	**BTK MUTATION: case report and current screening situation**	Tuan Minh Nguyen(Children Hospital No1)

APP3　APSID Poster-3

ENG

Saturday March 23, 2024　14:20-15:20 (Terrace Room)

Chair: Narissara Suratannon (Chulalongkorn University)
磯田 健志 /Takeshi Isoda (東京医科歯科大学 /Tokyo Medical and Dental University)

APP3-1	**A case of disseminated molluscum contagiosum in atopic triad with recurrent pneumonia and work-up of underlying combined immunodeficiency**	Carolyn Mae Matira Abelador (UP-Philippine General Hospital)

APP3-2	**Wiskott Aldrich Syndrome Suspected on a Critically-Ill 5 month-old Male**	Jemma Grace Fianza(Philippine General Hospital)
APP3-3	**Successful allogeneic hematopoietic stem cell transplantation for a male with EBV associated-hemophagocytic lymphocystiocytosis and X-linked Inhibitor of Apoptosis Deficiency: a Case Report in Taiwan**	Ching Yu Wang(National Taiwan University Hospital Yunlin Branch)
APP3-4	**TWO CASES OF COMBINED IMMUNODEFICIENCY CAUSED BY PTPRC MUTATIONS**	Yulu Li(Beijing Children's Hospital)
APP3-5	**HEMATOPOIETIC STEM CELL TRANSPLANTATION IN HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS: A SINGLE CENTRE EXPERIENCE**	Fouzia Nambiathayil Aboobacker(Christian Medical College Vellore)
APP3-7	**Spectrum of Systemic Autoinflammatory Diseases at a Tertiary Care Center in South India**	Neha Singh(ASTER CMI)

APP4　APSID Poster-4

ENG

Saturday March 23, 2024　14:20-15:20 (Terrace Room)

Chair: Wenwei Tu (Hong Kong University)
浅野 孝基 /Takaki Asano (広島大学 /Hiroshima University)

APP4-1	**Clinical courses and genetic analysis in Taiwanese patients with Syndromic diarrhea/Trichohepatoenteropathy (SD/THE)**	Wen-I Lee(Chang Gung University)
APP4-2	**Syndromic neurtropenia in Indian child**	Sathish Kumar Loganathan(Christian Medical College Vellore)
APP4-3	**Outcome for patients with severe congenital neutropenia treated outpatient granulocyte colony-stimulating factor at Vietnam National Children's Hospital**	Anh Phuong Ha(APSID)
APP4-4	**A Classic Case of Dyskeratosis Congenita**	Elaine Louise Lavilles Fernandez(Philippine General Hospital)
APP4-5	**Long road to recovery of one IFNGR1 deficiency case: from head to calf, from BCG to NTM**	Wenjing Zhang(Children's Hospital of Chongqing Medical University)

PL　Plenary Lecture

ENG

Saturday March 23, 2024　16:20-17:10 (Hall)　Sponsored by Miyarisan Pharmaceutical

Chair: 金兼 弘和 /Hirokazu Kanegane (東京医科歯科大学 /Tokyo Medical and Dental University)

PL	**Inborn errors of immunity as a cause of inflammatory bowel disease**	Holm Uhlig(University of Oxford)

SSY　共催シンポジウム　自己炎症性疾患における最新 Topics/ Sponsored Symposium　Latest Topics on Autoinflammatory Diseases

ENG

Saturday March 23, 2024　10:20-11:40 (Hall West)　Sponsored by Novartis Pharma

Chair: 西小森 隆太 /Ryuta Nishikomori (久留米大学 /Kurume University)

SSY-1	**Novel Autoinflammatory Disease Causal Gene Identification and Targeted Therapy**	Qing Zhou(Zhejiang University)
SSY-2	FMFの遺伝子型―表現型相関～我々の理解はどこまで進んだか～	本田 吉孝(京都大学)
	Current understanding of genotype-phenotype correlations in FMF - how far are we away from our real goal ?	Yoshitaka Honda(Kyoto University)

SSY-3 STING炎症シグナルを制御する分子基盤 田口 友彦(東北大学)
The molecular mechanism underlying the STING inflammatory signalling Tomohiko Taguchi(Tohoku University)

MS 共催モーニングセミナー /COVID-19 から学ぶ先天性免疫異常症 (IEI) と感染症対策
Sponsored Morning Seminar JPN

Saturday March 23, 2024 10:20-11:20 (Hall East) Sponsored by Astra Zeneca

Chair: 和田 泰三 /Taizo Wada (金沢大学 /Kanazawa University)

MS 先天性免疫異常症に学ぶ、COVID-19と闘う免疫系 今井 耕輔(防衛医科大学校)
Immune System fighting against COVID-19~Lessons Learned from Inborn errors of immunity~ Kohsuke Imai(National Defense Medical College)

LS1 ランチョンセミナー 1/Luncheon Seminar 1 JPN

Friday March 22, 2024 12:20-13:20 (Hall East) Sponsored by Japan Blood Products Organization

Chair: 大賀 正一 /Ohga Shouichi (九州大学 /Kyushu University)

LS1 原発性免疫不全症の診断と感染予防 高田 英俊(筑波大学)
Inborn Errors of Immunity: Diagnosis and Prevention of infectious diseases Hidetoshi Takada(University of Tsukuba)

LS2 ランチョンセミナー 2 家族性地中海熱 (FMF) 診断 / 治療最前線 / JPN
Luncheon Seminar 2 Latest Topics on how to diagnose and treat Familial Mediterranean fever(FMF)

Friday March 22, 2024 12:20-13:20 (Hall West) Sponsored by Novartis Pharma

Chair: 井田 弘明 /Hiroaki Ida (久留米大学 /Kurume University)

LS2-1 FMF 診断基準と自己炎症性疾患診療ガイドラインの改訂について 八角 高裕(京都大学)
Revised FMF Diagnostic Criteria and Guidelines for the Treatment of Autoinflammatory Diseases Takahiro Yasumi(Kyoto University)

LS2-2 家族性地中海熱と鑑別すべき疾患 桐野 洋平(横浜市立大学)
Diseases that should be differentiated from familial Mediterranean fever Yohei Kirino(Yokohama City University)

LS3 ランチョンセミナー 3/Luncheon Seminar 3 ENG

Saturday March 23, 2024 12:00-13:00 (Hall East) Sponsored by CSL Behring

Chair: 笹原 洋二 /Yoji Sasahara (東北大学 /Tohoku University)

LS3 **Immunoglobulin Therapy- From Mechanisms to Therapeutic Goals** Pamela Lee(Hong Kong University)

LS4　ランチョンセミナー 4　新生児期・乳児期に見逃してはいけない希少疾患 / Luncheon Seminar 4　Rare diseases not to be missed in newborns and infants　JPN

Saturday March 23, 2024　12:00-13:00 (Hall West)　Sponsored by Alexion Pharma

Chair: 伊達木 澄人 /Sumito Dateki (長崎大学病院 /Nagasaki University Hospital)

LS4-1	ライソゾーム酸性リパーゼ欠損症：血球貪食性リンパ組織球症(HLH)をきたす希少疾患	山田 勇気(大阪市立総合医療センター)
	Lysosomal acid lipase deficiency: Rare diseases presenting with hemophagocytic lymphohistiocytosis (HLH)	Yuki Yamada(Osaka City General Hospital)
LS4-2	小児難病の早期診断と治療	中村 公俊(熊本大学)
	Early diagnosis and treatment for intractable diseases in childhood	Kimitoshi Nakamura(Kumamoto University)

LS5　ランチョンセミナー 5/Luncheon Seminar 5　ENG

Sunday March 24, 2024　12:30-13:30 (Hall East)　Sponsored by Takeda Pharmaceutical

Chair: 岡田 賢 /Satoshi Okada (広島大学 /Hiroshima University)

LS5	**Children with rare diseases of the immune systems – from therapeutic orphans to pioneers of a new era of precision medicine**	Christoph Klein(Dr. von Hauners Children's Hospital)

LS6　ランチョンセミナー 6/Luncheon Seminar 6　ENG

Sunday March 24, 2024　12:30-13:30 (Hall West)　Sponsored by Pharming

Chair: 森尾 友宏 /Tomohiro Morio (東京医科歯科大学 /Tokyo Medical and Dental University)

Stuart Tangye (The Garvan Institute of Medical Research)

LS6-1	**Driving Discovery in Primary Immune Deficiency: Understanding APDS**	Stuart Tangye(The Garvan Institute of Medical Research)
LS6-2	**Driving Discovery in Primary Immune Deficiency: Understanding APDS**	森尾 友宏(東京医科歯科大学) Tomohiro Morio(Tokyo Medical and Dental University)

[IEI School 2024]

IEI1 IEI School Session 1 Antibody deficiency ENG

Friday March 22, 2024 8:40-9:40 (Room C)

Chair: 和田 泰三 /Taizo Wada (金沢大学 /Kanazawa University)
Huawei Mao (Beijing Children's Hospital)

IEI1-L	**Inborn Errors of Immunity (IEI) resulting in Primary Antibody Deficiencies (PAD)**	Hans D Ochs(University of Washington)
IEI1-1	**An infant girl with complex infections and agammaglobulinemia**	Nguyen Ngoc Tin(Children's Hospital No.1)
IEI1-2	**Disseminated molluscum contagiosum as the initial manifestation of Hyper IgE syndrome**	Isabella Ocampo Santos(Philippine General Hospital)
IEI1-3	**A case of a 10-year-old girl with NFKB2 deficiency who developed refractory thrombocytopenia treated by rituximab**	Hwanhee Park(Samsung Medical Center)

IEI2 IEI School Session 2 EI and TCR signaling defects ENG

Friday March 22, 2024 9:40-10:40 (Room C)

Chair: 岡田 賢 /Satoshi Okada (広島大学 /Hiroshima University)
Adli Bin Ali (Universiti Kebangsaan Malaysia)

IEI2-L	**T cell signaling defects in primary immune regulatory disorders (PIRD)**	Elena Hsieh(Colorado University)
IEI2-1	**X-linked anhidrotic ectodermal dysplasia with immunodeficiency: 14 years experience in a medical center in Taiwan**	Hsin-Hui Yu(National Taiwan University Children's Hospital)
IEI2-2	**A family with a potentially novel defect of a transcription factor gene affecting CD4 T cells**	Daniel Leung(The University of Hong Kong)
IEI2-3	**POLYAUTOIMMUNITY IN AN INFANT WITH LOW T-CELLS: AN INTERESTING CASE OF CD3ε DEFICIENCY AND REVIEW OF LITERATURE**	Neha Singh(Aster CMI Hospital)

IEI3 IEI School Session 3 IEI with autoimmune diseases ENG

Friday March 22, 2024 11:00-12:00 (Room C)

Chair: Woei Kang Liew (Mount Elizabeth Novena Hospital)
大西 秀典 /Hidenori Ohnishi (岐阜大学 /Gifu University)

IEI3-L	**Inborn Errors of Immunity; autoimmune disease**	Martin van Hagen(Erasmus University Medical Center)
IEI3-1	**Sirolimus Significantly Improves Symptoms in Patients with SOCS1 Haploinsufficiency**	Mengyue Deng(Beijing Children's Hospital)
IEI3-2	**Battling the staggering STATs: clinical heterogeneity of STAT1 gain-of-function mutations in a Chinese kindred**	Phoebe Qiaozhen Mak(Princess Margaret Hospital)
IEI3-3	**RAG1 deficiency with atypical severe combined immunodeficiency, autoimmunity, and gamma-delta T cells expansion in non-identical twins**	Pei-Jung Chung(National Taiwan University Children's Hospital)

IEI4　IEI School Session 4　Monogenic IBD　ENG

Friday March 22, 2024　15:00-16:00 (Room C)

Chair: Narissara Suratannon (Chulalongkorn University)
新井 勝大 /Katsuhiro Arai (国立成育医療研究センター /National Center for Child Health and Development)

IEI4-L	**The diagnostic approach to autoimmune enteropathies and monogenic IBD**	Holm Uhlig(University of Oxford)
IEI4-1	**Haploinsufficiency of A20 presenting as very-early-onset inflammatory bowel diseases and juvenile idiopathic arthritis**	Chester Huang(KK Women's and Children's Hospital)
IEI4-2	**Co-manifestation of Leukocyte Adhesion Deficiency Type II and Monogenic Inflammatory Bowel Disease Explained by Structural Modeling of SLC35C1 Variant**	Mahnaz Jamee(Willem-Alexander Children's Hospital)
IEI4-3	**A child with enteritis: Elucidating the importance of identifying the underlying monogenic etiology**	Sathish Kumar Loganathan(Christian Medical College Vellore)

IEI5　IEI School Session 5　IEI with cytopenia　ENG

Friday March 22, 2024　16:00-17:00 (Room C)

Chair: 笹原 洋二 /Yoji Sasahara (東北大学 /Tohoku University)
Yae-Jean Kim (Samsung Medical Center)

IEI5-L	**Autoimmune Cytopenias and Inborn Errors of Immunity: Recent Insights into Pathogenesis and Optimizing Therapy**	David B. Lewis(Stanford Univeristy)
IEI5-1	**A case of Wiskott-Aldrich syndrome successfully underwent allogeneic hematopoietic stem cell transplantation by treating immune thrombocytopenia with rituximab**	Saori Katayama(Tohoku University Hospital)
IEI5-2	**Mitochondrial disease with EARS2 mutation presenting with combined immunodeficiency: a case report**	Hsin-Ying Hsieh(National Taiwan University Children's Hospital)
IEI5-3	**De Novo deep intron *ELANE* mutation resulting in severe congenital neutropenia**	Zhou Shu(Beijing Children's Hospital)

IEIP-A　IEI Poster session-A　Ab, TCR, IBD, cytopenia　ENG

Friday March 22, 2024　13:30-15:00 (Terrace Room)

Chair: Jaime Sou Da Rosa Duque (Hong Kong University)
石毛 崇 /Takashi Ishige (群馬大学 /Gunma University)

IEIP-A-1	**A novel IKBKG intronic splicing mutation resulting in a completely loss of NEMO protein and causing a severe NEMO-ID phenotype**	Zhirui Tian(Children's Hospital of Chongqing Medical University)
IEIP-A-2	**A rare case of necrotizing encephalopathy in a CMV-infected boy with pancytopenia and abnormalities of skin, nails, and oral mucosa**	Tin Ngoc Nguyen(Children's Hospital No 1)
IEIP-A-3	**Severe combined immunodeficiency in CHARGE syndrome, the first case in Korea**	Luli Kim(Samsung Medical Center)
IEIP-A-5	**Tricho-Hepato-Enteric syndrome type 2: the first case report in Taiwan**	Jou-An Chen(National Taiwan University Children's Hospital)

IEIP-B IEI Poster session-B autoimmunity-1 ENG

Friday March 22, 2024 13:30-15:00 (Terrace Room)

Chair: Jonie Santos-Ocampo (Makati Medical Center and Asian Hospital)
金澤 伸雄 /Nobuo Kanazawa (兵庫医科大学 /Hyogo Medical University)

IEIP-B-2	**Refractory recurrent thrombocytopenia in 9-year-old boy**	Kanako Takeuchi(Hiroshima University)
IEIP-B-3	**Autoimmune Manifestations Prevalence among Chronic Cranulomatous Disease (CGD) Patients from a Primary Immunodeficiency Referral Center In Iran**	Samin Sharafian(Mofid Children's Hospital)
IEIP-B-5	**Nephrocalcinosis in Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy (APECED); Primary or Iatrogenic?**	Mohammadreza Shafiei(Alborz University)
IEIP-B-6	**An Adolescent Boy with Recurrent Painful Ulcer and Arthritis**	Diadra Annisa(Cipto Mangunkusumo Hospital)

IEIP-C IEI Poster session-C autoimmunity-2 ENG

Friday March 22, 2024 13:30-15:00 (Terrace Room)

Chair: Dina Muktiarti (Cipto Mangunkusumo Hospital)
磯田 健志 /Takeshi Isoda (東京医科歯科大学 /Tokyo Medical and Dental University)

IEIP-C-1	**Successful treatment of sirolimus for chronic diarrhea in LRBA Deficiency patient : A case report**	Natcharindhorn Thaweepolaungsuchawon (Ramathibodi Hospital)
IEIP-C-2	**An infant with recurrent fever with a somatic NRAS mutation**	Sophie Hon Yu Lai(Queen Mary Hospital)
IEIP-C-3	**Immunodysregulation and lymphoproliferation in a patient with PIK3CD despite sirolimus use**	Qin Ying Lim(Queen Mary Hospital)
IEIP-C-4	**Early onset autoimmunity in patients with Inborne errors of Immunity**	Madhubala Sharma(PGIMER)
IEIP-C-5	**Eight Cases of Autoinflammatory Recurrent Fevers**	Lan Shu(Women and Children's Hospital of Hubei Province)
IEIP-C-6	**Family analysis on case of Griscelli syndrome type 2**	Xi Yang(Children's Hospital of Chongqing Medical University)

IEIP-D IEI Poster session-D SCID, DADA2, NBS ENG

Friday March 22, 2024 13:30-15:00 (Terrace Room)

Chair: Wenwei Tu (Hong Kong University)
土居 岳彦 /Takehiko Doi (広島大学 /Hiroshima University)

IEIP-D-1	**Skin Hyperpigmentation as a new Presentation in 6 Patients with Adenosine Deaminase Deficiency(ADA)deficiency**	Samin Sharafian(Mofid Children's Hospital)
IEIP-D-2	**Demographic, clinical, immunological, and molecular features of iranian national cohort of patients with Artemis deficiency due to the defect in DCLRE1C gene**	Samin Sharafian(Mofid Children's Hospital)
IEIP-D-3	**Recurrent flare-associated urticaria in adenosine deaminase type 2 deficiency**	Pratap Kumar Kumar Patra(All India Institute of Medical Sciences)
IEIP-D-4	**Protean manifestations of *DADA2*: a physician's conundrum**	Prabal Barman(PGIMER)
IEIP-D-5	**Clinical and molecular profile of 20 patients with DOCK8 deficiency: A single-centre experience from Southern India**	Neha Singh(Aster CMI Hospital)
IEIP-D-6	**Newborn Screening for Primary Immunodeficiency Diseases (PID) in Malaysia: Current Status, Challenges and Progress**	Wai Leng Chang(Universiti Kebangsaan Malaysia)

IEIP-E IEI Poster session-E NFKB, STAT

ENG

Friday March 22, 2024 13:30-15:00 (Terrace Room)

Chair: Intan Hakimah Ismail (Universiti Putra Malaysia)

森谷 邦彦 /Kunihiko Moriya (防衛医科大学校 /National Defence Medical College)

IEIP-E-1	**Efficacy of tocilizumab in the treatment of *STAT3* GOF disease**	Meiping Lu(Zhejiang University Children's Hospital)
IEIP-E-2	**A Teenage Girl with Severe Tuberculosis Complicated with Aspergillosis and Candidiasis**	Fahreza Aditya Neldy(Universitas Indonesia)
IEIP-E-3	**Vast yet naïve. BENTA disease, an unusual presentation of extensive lymphoproliferation with *CARD11* gain-of-function mutation.**	Kent Mun Loh(KK Women's and Children's Hospital)
IEIP-E-4	**A unique clinical phenotype of *IKBKG* (nuclear factor-κB essential modulator) mutation presenting as severe central nervous system *Aspergillus terreus* infection**	Lin Lin Liu(Beijing Children's Hospital)
IEIP-E-5	**CARD9 deficiency with allergic bronchopulmonary aspergillosis (ABPA)-like presentation: a case report**	Mohammadreza Shafiei(Alborz University)

IEIP-F IEI Poster session-F CGD, LAD, general

ENG

Friday March 22, 2024 13:30-15:00 (Terrace Room)

Chair: Vignesh Pandiarajan (PGIMER)

山田 雅文 /Masafumi Yamada (酪農学園大学 /Rakuno Gakuen University)

IEIP-F-1	**INFANT WITH CHRONIC GRANULOMATOUS DISEASE ASSOCIATED INTESTINAL PERFORATION: A NEW ASSOCIATION?**	Rajkumar Kundavaram(All India Institute of Medical Sciences)
IEIP-F-2	**A zebra or a unicorn- a Chinese boy with McLeod Phenotype Chronic Granulomatous Disease**	Ki Crystal LAm(QMH)
IEIP-F-3	**An interesting case of CYBB mutation in a Malay boy with normal maternal DHR oxidation test and matched mother-child HLA haplotype**	Wan Fadhilah Wan Ibrahim(Universiti Putra Malaysia)
IEIP-F-4	**Targeted next-generation sequencing reveals a novel variant in *FERMT3* gene in a young girl with recurrent infections and epistaxis**	Amit Kumar(PGIMER)
IEIP-F-5	**Genomic approach to diagnose Primary Immunodeficiency Diseases (PIDs) in Bangladeshi Children**	Sudipta Roy(Ad-Din Medical College Hospital)
IEIP-F-6	**Paediatric Immunologists' Perspectives on Haematopoietic Stem Cell Transplantation (HSCT) for Primary Immunodeficiency Diseases (PID) in Malaysia**	Mohammad Shukri Khoo(The National University of Malaysia)