The 19th Korea-China-Japan Pediatric Nephrology Seminar 2023

The 2023 Spring Conference of the Korean Society of Pediatric Nephrology

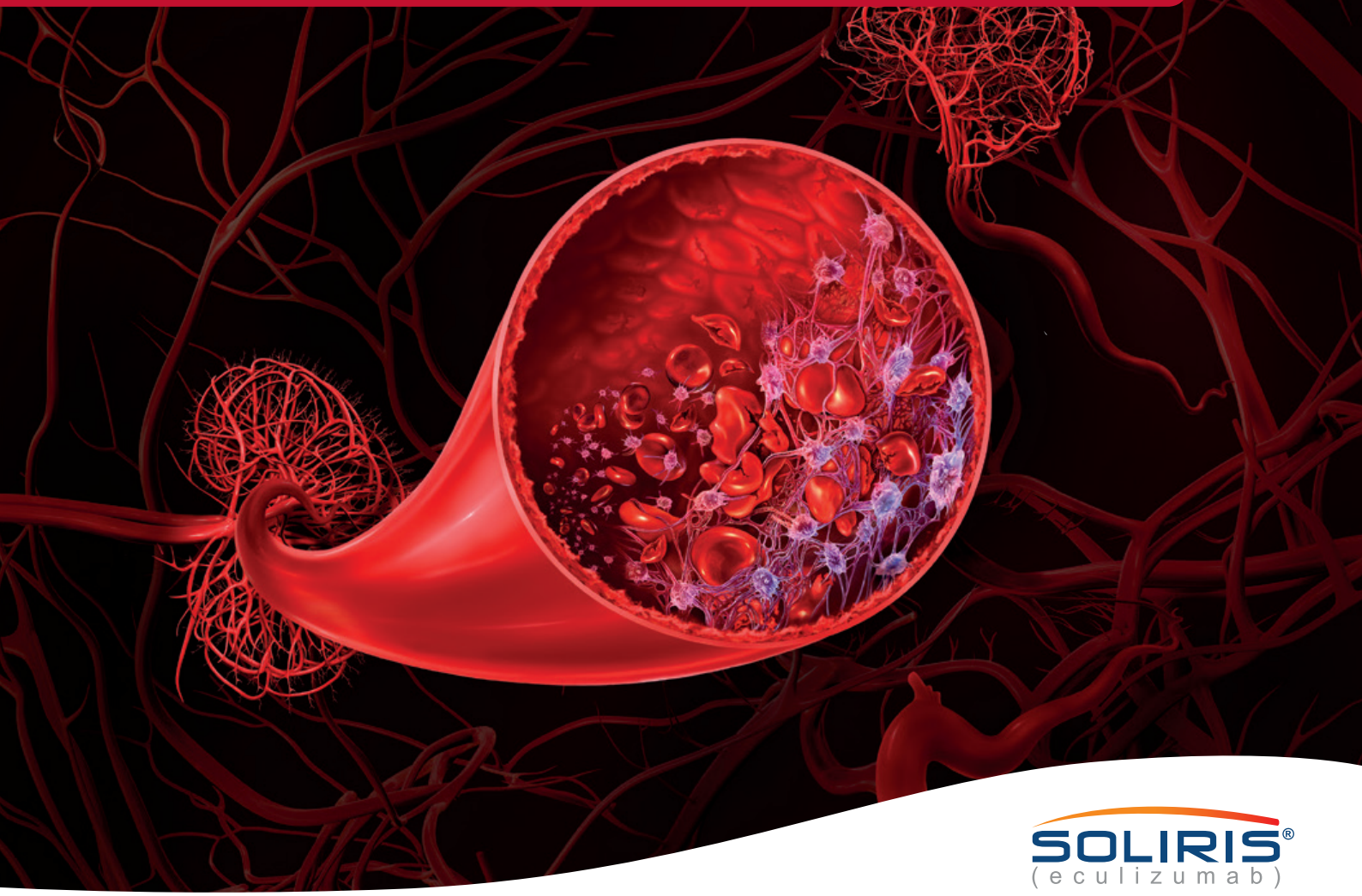
Date: 8th April (Sat), 2023

Format of seminar: Hybrid (Online & Offline)

Offline Venue: Auditorium(1F), Biomedical Research Institute,
Seoul National University Hospital, Seoul, Korea

Patients with aHUS can be at continuous risk of the life-threatening consequences of unpredictable complement-mediated TMA[1,2]

Chronic, uncontrolled complement activity in aHUS leads to ongoing endothelial injury, organ damage, and sudden death[2,3]



SOLIRIS® (eculizumab)

aHUS, atypical Hemolytic Uremic Syndrome; **TMA,** Thrombomicroangiopathy

References: 1. Laurence et al. Atypical Hemolytic Uremic; Essential Aspects of an Accurate Diagnosis. Clin Adv Hematol Oncol. 2016 Nov;14 Suppl 11(11)2-15. **2.** Legendre, C. M. et al. Terminal Complement Inhibitor Eculizumab N Engl J Med N Engl J Med 2013;368 2169-81. **3.** Noris et al. STEC HUS, atypical HUS and TTP are all, Nat. Rev. Nephrol. 2012 8, 622 633

prescribing information

전문의약품

솔리리스주(에쿨리주맙) [성분·함량] 1바이알(30mL)중 유효성분 : 에쿨리주맙(별규) 300mg 첨가제 : 염화나트륨, 인산수소나트륨 칠수화물, 인산이수소나트륨수화물, 주사용수, 폴리소르베이트80 **[효능·효과]** 1) 발작성 야간 혈색소뇨증(PNH : Paroxysmal Nocturnal Hemoglobinuria): 용혈을 감소시키기 위한 발작성 야간 혈색소뇨증(PNH : Paroxysmal Nocturnal Hemoglobinuria) 환자의 치료. 수혈 이력과 관계없이, 높은 질병 활성을 의미하는 임상 증상이 있는 환자의 용혈에 임상적 이익이 확립되었다. 2) 비정형 용혈성 요독 증후군(aHUS : atypical Hemolytic Uremic Syndrome): 보체 매개성 혈전성 미세혈관병증을 억제하기 위한 비정형 용혈성 요독 증후군(aHUS : atypical Hemolytic Uremic Syndrome) 환자의 치료 (사용제한: 시가(Shiga) 독신 생성 대장균에 의한 용혈성 요독 증후군(STEC-HUS) 환자 대상의 적용을 권장하지 않는다. 3) 시신경 척수염 범주 질환(Neuromyelitis optica spectrum disorder): 항아쿠아포린-4(AQP-4) 항체 양성인 환자의 시신경 척수염 범주 질환(NMOSD : Neuromyelitis optica spectrum disorder)의 치료 **[용법·용량]** 심각한 감염에 대한 위험을 줄이기 위해서 환자들은 최신의 백신 접종 지침(Advisory Committee on Immunization Practices(ACIP) recommendations)에 따라 백신 접종을 해야 한다.(사용상의 주의사항 1. 경고 항 참고) 이 약은 정맥투여되어야 하며 급속정맥투여(IV push) 또는 일시정맥투여(IV bolus)로 투여해서는 안된다. <성인> 1) 발작성 야간 혈색소뇨증(PNH) - 첫 4주간은 매 7일마다 600 mg을 투여한다. 네 번째 용량 투여 7일 후에 다섯 번째 용량으로 900 mg을 투여하고, 그 후부터는 매 14일마다 900 mg을 투여한다. 이 약은 권장 투여량과 일정에 맞게 투여, 혹은 예정된 일정의 2일 전/후로 투여 되어야 한다. 2) 비정형 용혈성 요독 증후군(aHUS) 및 시신경 척수염 범주질환(NMOSD) - 첫 4주간은 매 7일마다 900 mg을 투여한다. 네 번째 용량 투여 7일 후에 다섯 번째 용량으로 1200 mg을 투여하고, 그 후부터는 매 14일마다 1200 mg을 투여한다. <소아> 1) 비정형 용혈성 요독증후군(aHUS) 만 18세 미만의 aHUS 환자일 경우, 체중에 따라 다음의 일정으로 투여한다. [표 1] 만 18세 미만 환자에서의 권장 투여법 이 약은 권장 투여량과 일정에 맞게 투여, 혹은 예정된 일정의 2일 전/후로 투여되어야 한다. <혈장교환요법 및 신선 동결혈장투여사> 성인 및 소아 비정형 요독증후군과 성인 시신경 척수염 범주질환 환자에 대해 PE/PI(혈장 교환 요법(plasma exchange 또는 plasmapheresis), 또는 신선 동결 혈장 투여(fresh frozen plasma infusion))와 같은 부수적 시술을 받는 경우 추가 용량 투여가 필요하다.

환자 체중	초기 용량	유지 용량
40 kg 이상	4주간 매 7일마다 900 mg 투여	5주차에 1200 mg, 이후 매 14일마다 1200 mg 투여
30 kg이상 40 kg 미만	2주간 매 7일마다 600 mg 투여	3주차에 900 mg, 이후 매 14일마다 900 mg 투여
20 kg 이상 30 kg 미만	2주간 매 7일마다 600 mg 투여	3주차에 600 mg, 이후 매 14일마다 600 mg 투여
10 kg 이상 20 kg 미만	첫 주에 600 mg 투여	2주차에 300 mg, 이후 매 14일마다 300 mg 투여
5 kg 이상 10 kg 미만	첫 주에 300 mg 투여	2주차에 300 mg, 이후 매 21일마다 300 mg 투여

[표 2] PE/PI 이후 이 약의 추가적 투여법

부수적 시술의 종류	최근 사용한 이 약의 용량	부수적 시술 시 이 약의 추가 투여 용량	추가 투여 시점
혈장 교환 요법 (plasma exchange or plasmapheresis)	300 mg	혈장 교환 요법 시행시마다 300 mg씩	혈장 교환 요법 이후 60분 이내
	600 mg 또는 그 이상	혈장 교환 요법 시행시마다 600 mg씩	
신선 동결 혈장 투여 (fresh frozen plasma infusion)	300 mg 또는 그 이상	신선 동결 혈장 투여 시마다 300 mg씩	신선 동결 혈장 투여 60분 이전

[사용상의 주의사항] 1. 경고 : 중대한 수막구균 감염: 작용기전으로 인하여 이 약의 사용은 중대한 수막구균 감염(패혈증 그리고/또는 뇌수막염)에 대한 환자의 감수성을 증가시킨다. 이 약의 투여 환자에게서 치명적이고 생명을 위협하는 수막구균 감염이 발생하였다. 수막구균 감염은 어느 혈청군에 의해 서도 발생할 수 있지만, 이 약의 투여 환자들은 흔하지 않은 혈청군(X 등)에 의한 질환이 발생할 수 있다. 감염의 위험성을 낮추기 위하여, 이 약의 치료가 지연됨으로 인한 위험성이 수막구균 감염 발생의 위험성보다 큰 경우를 제외하고는 모든 환자들은 반드시 이 약의 투여 시작 최소한 2주 전에 수막구균 백신을 투여 받아야 한다. 만약 접종 받지 않은 환자가 긴급히 이 약의 치료를 받아야 하면, 최대한 빨리 수막구균 백신을 투여 받도록 한다. 수막구균 백신 접종 이후 2주 이내 이 약을 투여할 경우, 4가 수막구균 백신 접종 이후 2주 동안 적절한 예방적 항생요법으로 치료 받아야 한다. 흔한 병원성 수막구균 혈청군을 예방하기 위하여 가능하다면 혈청군 A, C, Y, W135, B에 대한 백신이 권장된다. 환자들은 백신 사용을 위한 최신의 백신 접종 지침(Advisory Committee on Immunization Practices(ACIP) recommendations)에 따라 백신을 접종 혹은 재접종 받아야 한다. 백신 접종은 보체를 더욱 활성화시킬 수 있다. 결과적으로, PNH, aHUS, 불응성 gMG 및 NMOSD를 포함한 보체 매개 질환을 가진 환자들은 용혈(PNH의 경우)이나 혈전성 미세혈관병증(TMA; aHUS의 경우) 또는 중증 근무력증의 악화(불응성 gMG의 경우)또는 재발(NMOSD의 경우)과 같은 그들의 기저 질환의 징후 및 증상이 증가하는 경험을 할 수 있다. 따라서, 지침에 따른 백신 접종 이후 질환의 증상에 대해 면밀히 관찰되어야 한다. 백신 접종은 수막구균 감염 위험을 줄일 수 있지만, 완전히 없애지는 않는다. 적절한 항생제 사용에 대한 공식 지침(예: 국내 성인 세균성 수막염의 임상진료지침 권고안 등)을 고려하여야 한다. 수막구균 감염의 초기 징후나 증상이 나타나는지 면밀히 관찰하고, 감염이 의심되면 즉시 검사받아야 한다. 환자는 이러한 징후와 증상 및 즉시 치료를 받는 절차에 대해 안내 받아야 하며, 담당 의사는 반드시 환자와 이 약의 치료의 위험과 이익을 상의하여야 한다. 수막구균 감염은 초기에 발견하고 치료하지 않으면 급격히 치명적이고 생명을 위협하게 될 수 있다. 중대한 수막구균 감염을 치료받는 환자는 이 약의 투여를 중지하도록 한다. 2. 다음 환자에는 투여하지 말 것 1) 이 약의 주성분, 뮤린 단백질 또는 기타 구성성분에 과민반응이 있는 환자 2) 치료되지 않은 중대한 수막구균(Neisseria meningitidis) 감염 환자 3) 수막구균(Neisseria meningitidis) 백신을 현재 접종하지 않은 환자 또는 백신 접종 이후 2주 동안 적절한 예방적 항생요법으로 치료를 받지 않은 환자(이 약의 치료를 늦추는 것이 수막구균 감염을 일으키는 것보다 중대하지 않은 경우) **[수입자,허가권]** (주) 한독서울특별시 강남구 테헤란로 132전화번호: (02) 527-5114자 개정년월일: 2023년 02월 03일 **※ 보다 자세한 사항은 제품설명서 전문을 참고하시기 바랍니다.** aSOL20230206

KR-13009 I Exp.2025-02(Prep.2023-02)

ALEXION

한국아스트라제네카 서울 강남구 영동대로 517 아셈타워 21층 전화: (02)2188-0800 팩스:(02)2188-0852

AstraZeneca

▸▸ Welcome Message

Dear Friends and Colleagues,

Welcome to the 19th Korea-China-Japan Pediatric Nephrology Seminar!

The 19th Korea-China-Japan Pediatric Nephrology Seminar was started for scientific communication in pediatric nephrology as well as friendship among pediatric nephrologists of each country.

Unfortunately, in 2022, it was not held due to the corona pandemic, but on April 8, 2023, we are pleased to be able to host this seminar in Seoul in a hybrid format that combines face-to-face and online. We hope that this seminar will serve as an opportunity for a new advancement in pediatric nephrology and many of you will be able to enjoy the beautiful spring days together in Seoul.

Sincerely yours,

Jihong Kim

Ji-Hong Kim, M.D., Ph.D.
President
The Korean Society of Pediatric Nephrology

▸▸ Organization

KOREA

Hae Il Cheong	Seoul National University
Il-Soo Ha	Seoul National University
Hee Gyung Kang	Seoul National University
Kee Hyuck Kim	NHIS Ilsan Hospital
Joo Hoon Lee	University of Ulsan, Asan Medical Center
Young Seo Park	University of Ulsan, Asan Medical Center
Kee-Hwan Yoo	Korea University
Min Hyun Cho	Kyungpook National University (Liaison officer)
Ji Hong Kim	Yonsei University
Yong Jin Kim	Kyungpook National University, School of Medicine
Tae-Sun Ha	Chungbuk National University
Ja Wook Koo	Inje University
Jae Il Shin	Yonsei University

CHINA

Jie Ding	Peking University First Hospital
Xiaoyun Jiang	The First Affiliated Hospital of Sun Yat-sen University
Jianhua Mao	The Children's Hospital, Zhejiang University School of Medicine
Fang Wang	Peking University First Hospital
Suxia Wang	Peking University First Hospital
Zhuwen Yi	The Second Xiangya Hospital of Central South University
Aihua Zhang	Nanjing Children's Hospital
Xuhui Zhong	Peking University First Hospital (Liaison officer)
Jianhua Zhou	Tongji Hospital, Huazhong University of Science and Technology
Zhengkun Xia	Jinling Hospital
Qian Shen	Children's Hospital of Fudan University
Songming Huang	Children's hospital of Nanjing Medical University
Qiu Li	Children's Hospital of Chongqing Medical University
Hong Xu	Children's Hospital of Fudan University

JAPAN

Kunimasa Yan	Kosei Hospital
Kazumoto Iijima	Hyogo Prefectural Kobe Children's Hospital
Michio Nagata	Diagnostic Pathology, Itabashi Chuo Medical Center
Motoshi Hattori	Department of Pediatric Nephrology, Tokyo Women's Medical University
Koichi Nakanishi	Department of Child Health and Welfare (Pediatrics), Graduate School of Medicine, University of the Ryukyus
Yoshitsugu Kaku	Nephrology Fukuoka Children's Hospital
Shuichi Ito	Department of Pediatrics, Yokohama City University Graduate School of Medicine
Keisuke Sugimoto	Department of Pediatrics, Kindai University Faculty of Medicine
Rika Fujimaru	Department of Pediatrics, Osaka City General Hospital
Yuko Shima	Department of Pediatrics, Wakayama Medical University
Ryugo Hiramoto	Children's Medical Centre, Matsudo City General Hospital (liaison)

▸▸ General Information

MEETING DATE AND VENUES

The 19th Korea-China-Japan Pediatric Nephrology Seminar 2023 &
The 2023 Spring Conference of the Korean Society of Pediatric Nephrology

Date: April 8 (Sat), 2023
Format of seminar: Hybrid (Online & Offline)
Offline Venue: Auditorium(1F), Biomedical Research Institute, Seoul National University Hospital, Seoul, Korea

WEB SEMINAR

The 19th Korea-China-Japan Pediatric Nephrology Seminar 2023 will be conducted both online and offline simultaneously as a hybrid conference.

- All people attending the event must register in advance on the conference website.
- Only pre-registered applicants can attend the Online Web Seminar.
- Due to the nature of Online Web Seminar, same-day registration is not possible.
- When registering, please be sure to enter your name and e-mail address as they are required for login.
- Web Seminar link: https://kspn.org/register/2023_01/webinar/info.html

ACCOMMODATION

The Korean Society of Pediatric Nephrology has assigned the 'ORAKAI Daehakro Hotel
Address: 180, Yulgok-ro, Jongno-gu, Seoul, 03127, Republic of Korea
Contact Information:
T +82 2 6353 7700 / E rsvn.daehakro@orakaihotels.com / H https://dh.orakaihotels.com/eng/default.asp

ORGANIZING COMMITTEE MEETING

Date: Friday 7, April, 18:00 (6:00 PM)

Venue (Business meeting & dinner) : Samcheonggak (Cheonchudang, Seoul)

REGISTRATION

- Online registration only, on-site registration is unavailable.
- Pre-registration link: https://kspn.org/register/2023_01/entry/info.html
- Individual registrations only, group registrations will not be accepted.
- Only Korean participants will pay the registration fee.

CLOSING DINNER

Banquet will be served for all the attendees after the meeting ends.

Date: 17:30 on April 8, 2023

Dinner Place: Arirang

CONTINUOUS PROFESSIONAL DEVELOPMENT GUIDELINES

- Total 4 CPD lectures, 2 from hosting country, 1 from each participating country
- Official language: English
- Presentation 25min, Q&A 5min, total 30min

ORAL PRESENTATION GUIDELINES

- Total 9 Oral Presentation
- Official language: English
- Presentation 20min, Q&A 5min, total 25min
- Online Presentation & Offline Presentation

▬ Online Presentation

- Online presenters will be presenting through pre-recorded video and Q&A will be done live through ZOOM, the video conference program.
  ▷ It should be below 20 minutes.
  ▷ Additional 5-minute Q&A will be done live using the ZOOM program.

▸▸ General Information

▷ ZOOM link: https://us06web.zoom.us/j/7202687795?pwd=L1FqWm5sVkkzdDlhSm9wMmk1d3Jtdz09

▬ Offline Lecture Presentation

- Offline participants don't need to register for pre-recorded lecture videos, but register lecture slides in the pre-view room when you come to the seminar and present a lecture.

POSTER ORAL PRESENTATION GUIDELINES

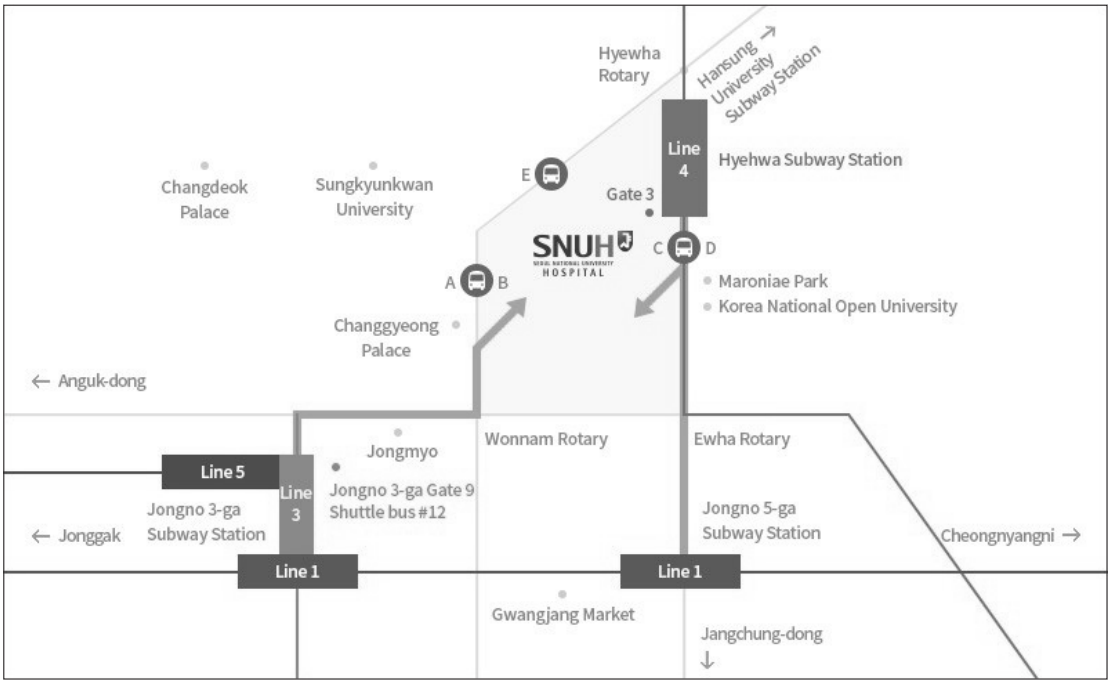
- Total 12 Poster Oral Presentation
  ▷ All Offline Presentation
- Official language: English
- 5 minutes for each presentation, 3 minutes for discussion.
- Presentation Location: Lobby of the seminar venue
- Presentation Method: E-poster presentation using DID

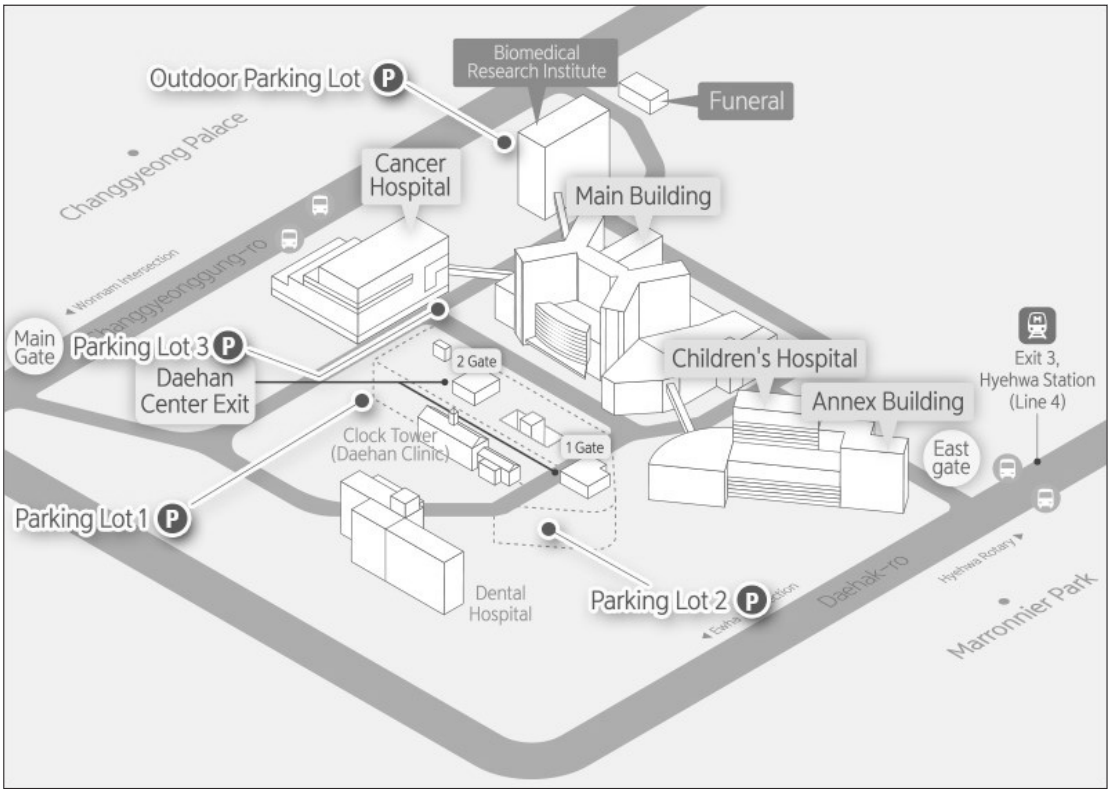
POSTER EXHIBITION GUIDELINES

- Poster exhibition will be done through the online website and onsite DID without additional onsite presentation.
- Total 11 Poster Exhibition
- Official language: English

TRANSPORTATION

- Address: Seoul National University Hospital Biomedical Research Institute 101 Daehak-ro Jongno-gu, Seoul 100-744 South Korea


Hyewha Rotary
Hansung University Subway Station
Line 4
Hyehwa Subway Station
Changdeok Palace
Sungkyunkwan University
E
Gate 3
SNUH
C
D
A
B
Maroniae Park
Korea National Open University
Changgyeong Palace
← Anguk-dong
Jongmyo
Wonnam Rotary
Ewha Rotary
Line 5
Line 3
Jongno 3-ga Gate 9
Shuttle bus #12
Jongno 3-ga Subway Station
Jongno 5-ga Subway Station
← Jonggak
Cheongnyangni →
Line 1
Line 1
Gwangjang Market
Jangchung-dong
↓


Outdoor Parking Lot
Biomedical Research Institute
Funeral
Changgyeong Palace
Cancer Hospital
Main Building
Changgyeonggung-ro
Wonnam Intersection
Main Gate
Parking Lot 3
Daehan Center Exit
2 Gate
Children's Hospital
Annex Building
Exit 3, Hyehwa Station (Line 4)
Clock Tower (Daehan Clinic)
1 Gate
East gate
Parking Lot 1
Dental Hospital
Parking Lot 2
Daehak-ro
Hyehwa Rotary
Marronnier Park

▸▸ General Information

SUBWAY

From -Seoul Station	4 Seoul Station Line No.4 → Hyehwa Station Exit. 3
From Seoul Express Bus Terminal	3 Express Bus Terminal Line No.3 → 4 Choongmuro Station (Transfer Line No.4) → Hyehwa Station Exit. 3
From Seoul Nambu Terminal	3 Nambu Bus Terminal Line No.3 → 4 Choongmuro Station (Transfer Line No.4) → Hyehwa Station Exit. 3
From Dongseoul Bus Terminal	2 Gangbyeon Station. Line No.2 → 4 Dongdaemun Stadium Station (Transfer Line No.4) → Hyehwa Station Exit. 3
From Sangbong Bus Terminal	7 Sangbong Station. Line No.7 → 5 Gunja Station.(Transfer Line No.5) → 4 Dongdaemun Stadium Station (Transfer Line No.4) → Hyehwa Station Exit. 3
From Gimpo Airport	5 Gimpo Int'l Airport Station. Line No.5 → 4 Dongdaemun Stadium Station (Transfer Line No.4) → Hyehwa Station Exit. 3

BUS

Changkyunggung down Platform (#A)	Blue	Blue 100, 102, 104, 106, 107, 108, 140, 143, 149, 150, 151, 160, 162, 171, 172, 272, 301, 710
	Airport	Airport 6011 (Tour Bus : 90S)
Changkyunggung up Platform (#B)	Blue	Blue 151, 171, 172, 272, 601
Daehangno Hyehwa Station Exit. 3 A down Platform (#C)	Blue	Blue 109, 273, 601, N16
	Green	2112, Jongro 07, Jongro 08, Jongro 12 (around Hospital)
Daehangno Hyehwa Station Exit. 3 A up Platform (#D)	Blue	Blue 100, 102, 104, 106, 107, 108, 109, 140, 143, 150, 160, 162, 273, 301, 710, N16
	Green	2112, Jongro 07, Jongro 08 (Tour Bus : 90S)

Airport(Bus & Subway)

1. Bus(#E)

Route Name (Bus number)	Airport → Changgyeonggung (Palace) / Seoul National University Hospital (6011)
Bus Type	Limousine (Standard)
Direction	Seoul(North)
Station	World Cup Stadium → Seongsan Hall → Yeonhui Sageori → Seodaemun-gu Office → Grand Hilton Hotel → Hongje Station → Muakjae → Gyeongbokgung station → Anguk-dong Stop → Changdeokgung → Sungkyunkwan Univ. → Hansung Univ. Station → Sungshin Women's Univ. Station
Bus Stop No.	Incheon International Airport (T1): 5B-3 platform Incheon International Airport (T2): 31 platform

Airport Bus Info

- Interval : Every 20~30 min.
- Traveling Time : 68~73 min
- First Bus: From Airport - 05:40 / From Chabggyeonggung (Palace) - 04:20
- Last Bus: From Airport - 22:40 / From Chabggyeonggung (Palace) - 22:40
- Fee : 10,000 won
- Vendor Name : Airport Limousine
- Contact No. : 82-2-2664-9898
- Homepage Information : http://www.airportlimousine.co.kr/

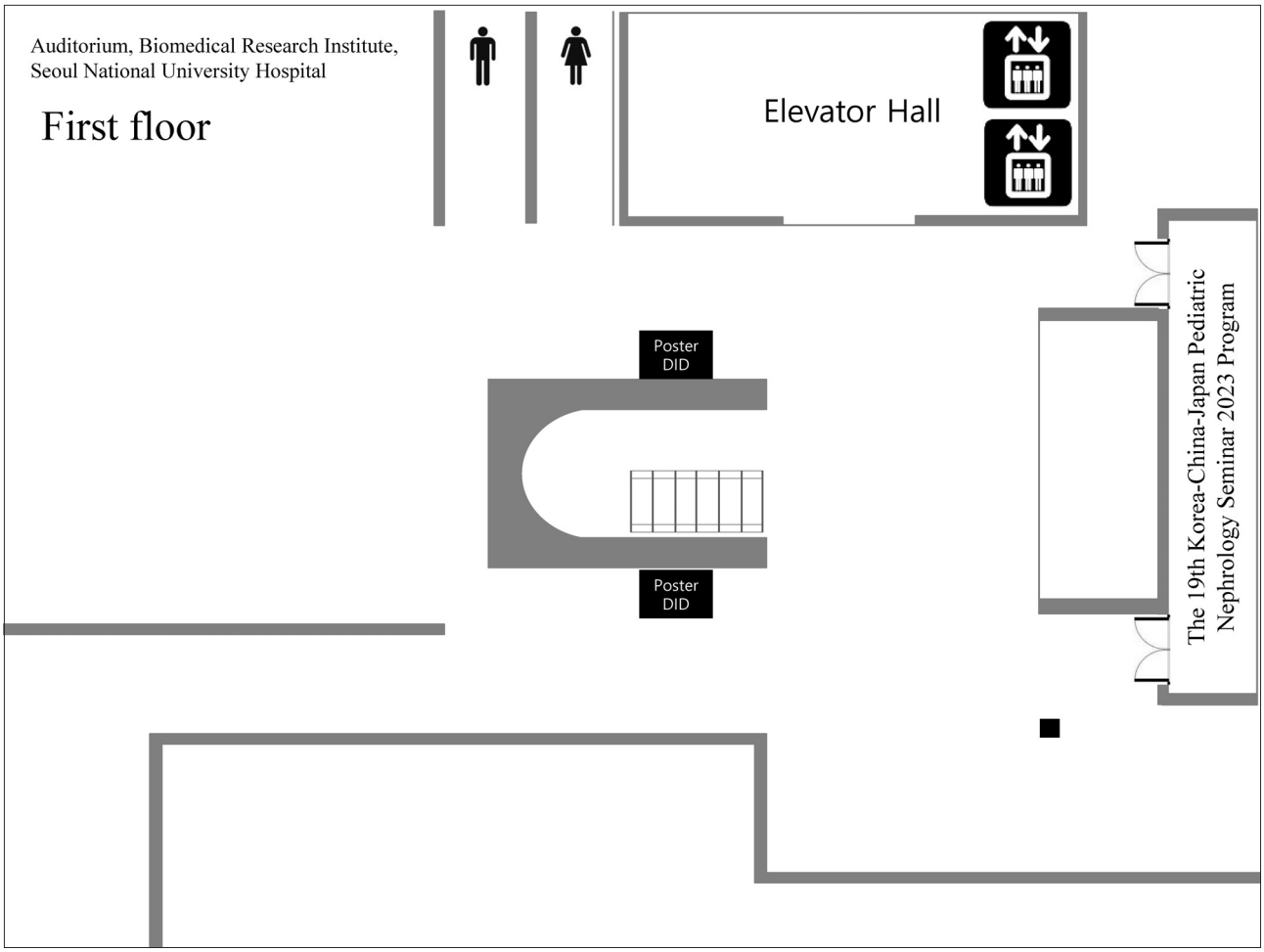
2. Subway

From Gimpo Airport	5 Gimpo Int'l Airport Station. Line No.5 → 4 Dongdaemun Stadium Station (Transfer Line No.4) → Hyehwa Station Exit. 3
From Incheon International Airport	Incheon Rapid Transit → 5 Gimpo Int'l Airport Station. Line No.5 → 4 Dongdaemun Stadium Station.(Transfer Line No.4) → Hyehwa Station Exit. 3

Car(Navigation Search)

Title Search	Seoul National University Hospital (서울대학교병원)
Address Search	101 DAEHAK-RO JONGNO-GU, SEOUL (서울시 종로구 대학로 101, 연건동)

▸▸ Floor Guide


Auditorium, Biomedical Research Institute,
Seoul National University Hospital
First floor
Elevator Hall
Poster
DID
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The 19th Korea-China-Japan Pediatric
Nephrology Seminar 2023 Program

Program

The 19th Korea-China-Japan Pediatric Nephrology Seminar 2023 & The 2023 Spring Conference of the Korean Society of Pediatric Nephrology

- **Date:** April 8 (Sat), 2023
- **Format of seminar:** Hybrid (Online & Offline)
- **Offline Venue:** Auditorium(1F), Biomedical Research Institute, Seoul National University Hospital, Seoul, Korea

08:30-08:55	Registration	
08:55-09:00	Opening Remark	Ji-Hong Kim (President of the Korean Society of Pediatric Nephrology)

09:00-09:30 Continuous Professional Development 1

Chairperson: Heeyeon Cho (Samsung Medical Center, Sungkyunkwan University, Korea)

09:00-09:25	National and international guidelines and clinical practice recommendations of Nephrotic Syndrome	Xuhui Zhong (Peking Unversity First Hospital, China)
09:25-09:30	Q&A	

09:30-09:55 Oral Presentation 1

Chairperson: Xiaoyun Jiang (The First Affiliated Hospital, Sun Yat-sen University, China)

09:30-09:50	Refractory nephrotic syndrome with prolonged hypogammaglobulinemia after rituximab treatment - A case exhibiting NFKB1 variant linked with common variable immunodeficiency	Miyu Sai (Tohoku University Graduate School of Medicine, Japan)
09:50-09:55	Q&A	

09:55-10:20 Oral Presentation 2

Chairperson: Rika Fujimaru (Osaka City General Hospital, Japan)

09:55-10:15	Persistent benign proteinuria associated with CUBN variants	Yun Young Choi (Seoul National University Children's Hospital, Korea)
10:15-10:20	Q&A	

10:20-10:45	**Oral Presentation 3**
	Chairperson: Min Hyun Cho (Kyungpook National University Hospital, Korea)
10:20-10:40	A Novel MYH9 Mutation in a Chinese Child with Immune Complex-Mediated Glomerulonephritis and Literature Review Jing Song (Children's Hospital of Chongqing Medical University, China)
10:40-10:45	Q&A
10:45-11:00	Break

11:00-11:30	**Continuous Professional Development 2**
	Chairperson: Ryugo Hiramoto (Children's Medical Centre, Matsudo City General Hospital)
11:00-11:25	Approach to microscopic hematuria in children Eujin Park (Korea University Guro Hospital, Korea)
11:25-11:30	Q&A

11:30-11:55	**Oral Presentation 4**
	Chairperson: Hyung Eun Yim (Korea University Ansan Hospital, Korea)
11:30-11:50	Multinucleated podocyte may lead to a diagnosis of cystinosis Ayako Ogata (Saiseikai Yokohamashi Nanbu Hospital, Japan)
11:50-11:55	Q&A

11:55-12:20	**Oral Presentation 5**
	Chairperson: Fang Wang (Peking Unversity First Hospital, China)
11:55-12:15	Refractory hyperkalemia after kidney transplantation in a Korean pediatric patient Jeong Yeon Kim (Chungnam National University Hospital, Korea)
12:15-12:20	Q&A
12:20-13:20	Luncheon

▸▸ Program

13:20-14:20	**Poster Presentation with Chair**
	Chairperson: Seong Heon Kim (Seoul National University Children's Hospital, Korea), Eunmi Yang (Chonnam National Univ., Korea)

14:20-14:45	**Oral Presentation 6**
	Chairperson: Jung Won Lee (Ewha Womans University Medical Center, Korea)
14:20-14:40	An 8 month old boy with infantile nephrotic syndrome caused by semaphorin 3B associated membranous nephropathy Yu Tanaka (Kobe University Graduate School of Medicine, Kobe, Japan)
14:40-14:45	Q&A

14:45-15:10	**Oral Presentation 7**
	Chairperson: Yasuko Kobayashi (Gunma University, Japan)
14:45-15:05	Single-cell transcriptomics reveal a B cells-mediated immune mechanism of the onset of immunoglobulin-A vasculitis nephritis: gut-homing B cells migrate to the kidney? Yaping Liu (Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, China)
15:05-15:10	Q&A

15:10-15:40	**Continuous Professional Development 3**
	Chairperson: Jin-Soon Suh (Bucheon St. Mary's Hospital, The Catholic University of Korea)
15:10-15:35	National and international guidelines and clinical practice recommendations of IgA nephropathy Yuko Shima (Wakayama Medical University, Japan)
15:35-15:40	Q&A
15:40-16:00	Break

16:00-16:25	**Oral Presentation 8**
	Chairperson: Jianhua Zhou (Tongji Hospital.Tongji Medical College, Huazhong University of Science and Technology, China)
16:00-16:20	Renal complications of Pediatric Glycogen Storage Disease, type I Min Ji Park (Kyungpook National University Hospital, Korea)
16:20-16:25	Q&A

16:25-16:50	Oral Presentation 9
	Chairperson: Shuichi Ito (Yokohama City University Graduate School of Medicine, Japan)
16:25-16:45	A Case of Thrombotic Microangiopathy Caused by Hypereosinophilic Syndrome Xiaoyu Liu, Yali Ren (Peking Unversity First Hospital, China)
16:45-16:50	Q&A

16:50-17:20	Continuous Professional Development 4
	Chairperson: Jianhua Mao (The Children Hospital of Zhejiang University School of Medicine, China)
16:50-17:15	Atypical Hemolytic Uremic Syndrome Hee Gyung Kang (Seoul National University Children's Hospital, Korea)
17:15-17:20	Q&A

▸▸ Contents

Continuous Professional Development

Oral Presentation

Poster Presentation

▸▸ Contents

▸▸ Contents

Continuous Professional Development

National and international guidelines and clinical practice recommendations for childhood nephrotic syndrome

Xuhui Zhong

Peking Unversity First Hospital, China

Idiopathic nephrotic syndrome is one of the most prevalent glomerulopathies in children. It is characterized by heavy proteinuria, hypoalbuminemia, edema, and hypercholesteremia. The incidence of childhood nephrotic syndrome varies among countries, ranging from 1.15 to 16.9 per 100,000 children annually. Compared with Europeans, South Asian children are reported to have a higher incidence of nephrotic syndrome. Whereas children from South and East/Southeast Asia have significantly lower probabilities of frequent relapses. During the past years, multiple international and national guidelines have been established or updated. The guidelines vary in the definition, classifications, and immunosuppressive treatment of childhood nephrotic syndrome. The prescription of immunosuppressive strategy is guided by classification, disease severity, adverse effects, cost of therapy, etc. We aim to review and compare recent international and national clinical practice guidelines for idiopathic nephrotic syndrome in children. Although nephrotic syndrome is relatively common in clinical practice, there are still unsolved questions in disease classification, appropriate duration of corticosteroids, and the choice of immunosuppressive agents.

Approach to microscopic hematuria in children

Eujin Park

Department of Pediatrics, Korea University Guro Hospital

Hematuria, defined as five or more red blood cells per high power field, is one of the common urine abnormalities that school-age children encounter in clinical practice. Even if the urine is red in color or the urine dipstick detects urinary occult blood, the urine may not contain RBCs; therefore, microscopic examination is required to confirm hematuria. The causes of hematuria in children range from infections to hypercalciuria, nephrolithiasis, vascular abnormalities, including nutcracker syndrome, acute and chronic glomerular diseases, urinary tract malformations, and tumors, and the differential diagnosis depends on whether the hematuria is glomerular or non-glomerular, and on the presence of comorbidities. Asymptomatic microscopic hematuria has clinical significance when it is persistent, so if it persists, a differential diagnosis of the cause is necessary. Persistent microscopic hematuria in children and adolescents should be differentiated through careful history taking, physical examination, blood and urine tests, and imaging studies, if indicated. Several international guidelines have been proposed for the diagnosis, evaluation, and follow-up of microscopic hematuria in adults, but there are only a few guidelines for children. In this talk, we will review guidelines on the evaluation and management of asymptomatic hematuria in children from: the Japanese guidelines of the management of hematuria 2013; the ACR Appropriateness Criteria Hematuria-Child 2018; and the Korean society of pediatric nephrology 2023 recommendations on diagnosis and management of children with hematuria.

National and international guidelines and clinical practice recommendations of IgA nephropathy

Yuko Shima, Koichi Nakanishi

Department of Pediatrics, Wakayama Medical University, Department of Child health and Welfare (Pediatrics), Graduate School of Medicine, University of Ryukyus

IgA Nephropathy (IgAN) is the most common primary glomerulonephritis in all ages in the world. The prevalence of IgAN displays remarkable variation in different parts of the world and across different ethnic groups. Some of this variability can be attributed to a racial difference due to the genetic susceptibility and or to differences in biopsy selection practices. In Japan, about 70% of the pediatric IgAN patients are found with asymptomatic hematuria and/or proteinuria by an annual school screening program since 1974 unlike those discovered with macroscopic hematuria in Europe and the United States.

Initially it seemed to be asymptomatic and had a good clinical course, 30 to 40% of patients reached ESKD in 20 years without treatments. And establishment of appropriate treatments for pediatric IgAN was desired in Japan. Then the Japanese Pediatric IgAN Treatment Study Group involving 20 Japanese pediatric renal centers have conducted several clinical trials since 1990, and shown that treatments in the early stage of the disease can prevent the disease progression. Now we use 2 years of combination therapy including prednisolone, mizoribine, and lisinopril for patients with severe childhood IgAN showing diffuse mesangial proliferation (WHO) and 2 years of lisinopril for mild cases according to the treatment guidelines 2020 for childhood IgAN by the Japanese society for pediatric nephrology. However, the current status of treatments for IgAN in Japan is unique compared to the internationally widely used KDIGO clinical practice guidelines because the summation of global evidence does not support the Japanese treatment policy. However, we need each compromise plan tailored to the clinical situation of each country following the international guideline as much as possible with the understanding that international guidelines prescribe treatment based on the accumulation of evidence in the countries where there is no school screening program and medical systems are different.

Atypical Hemolytic Uremic Syndrome

Hee Gyung Kang

Seoul National University College of Medicine, Seoul, Korea

In children, hemolytic uremic syndrome (HUS) is a common cause of acute kidney injury (AKI). It is characterized by microangiopathic hemolytic anemia, thrombocytopenia, and AKI. Although diarrhea-associated typical HUS is the prevalent type, some cases of HUS are caused by dysregulation of the complement alternative pathway, and these cases are denoted as atypical HUS (aHUS) or complement-mediated thrombotic microangiopathy (CM-TMA). Unlike typical HUS, aHUS often recurs and has a poor prognosis. However, the recent introduction of complement inhibition therapy as a targeted treatment has changed the management paradigm of this condition. In line with this advancement, national and international (by the international pediatric nephrology association) guidelines or clinical practice recommendations (CPR) have been published or are under development.

The Korean CPR, published in 2016, states that delayed diagnosis and treatment of aHUS can lead to death or end-stage kidney disease. Therefore, it is crucial to consider the possibility of aHUS in all patients who exhibit TMA. Since aHUS or CM-TMA is a diagnosis of exclusion, other causes of TMA, including severe deficiency of ADAMTS13 (thrombocytopenic thrombotic purpura), Shigatoxin-induced HUS, and secondary causes of TMA (infections, drugs, malignant hypertension, autoimmune diseases, and others) need to be ruled before diagnosing a patient with aHUS or CM-TMA. However, some cases of secondary TMA may later turn out to be aHUS, especially those with malignant hypertension or refractory cases.

In children with CM-TMA, the treatment of choice is complement inhibitor therapy, and early introduction of this therapy is associated with a better outcome. On the other hand, the benefit of plasma therapy is uncertain, except in cases with anti-factor H antibodies, where plasma exchange is indicated. Since aHUS is an episodic disease that may recur after triggering events such as intercurrent infections, discontinuation of complement inhibitor therapy can be considered after resolution of the

aHUS episode, except in cases with proven pathogenic genetic variants of the complement system, recurrent diseases, or kidney transplantation. In factor H antibody-mediated aHUS, immunosuppressive therapy with prednisone and anti-proliferative agents is indicated. When it comes to kidney transplantation, the recurrence of aHUS needs to be considered, and prophylactic complement inhibitor therapy is required in recurrent disease.

Oral Presentation

O-1

Refractory nephrotic syndrome with prolonged hypogammaglobulinemia after rituximab treatment - A case exhibiting NFKB1 variant linked with common variable immunodeficiency

Miyu Sai, Nao Uchida, Noriko Sugawara, Masahiro Irie, Yoji Sasahara, Atsuo Kikuchi

Department of Pediatrics, Tohoku University, Japan

Backgrounds: Rituximab (RTX) has been proven to be effective for refractosry nephrotic syndrome; however, persistent hypogammaglobulinemia is occasionally observed in some patients. This is generally regarded as a side effect of repeated RTX administra tion. We report a case in which a genetic analysis was performed in consideration of the coexistence of common variable immunodeficiency (CVID).

Case: A 13-year-old girl who developed nephrotic syndrome at the age of 2 was treated with a total of 8 cycles of rituximab for refractory nephrotic syndrome from the age of 5, after which, her CD19 cells depleted for about 38 months. Subsequently, she was able to discontinue immunosuppressive agents, including steroids, and has been in remission ever since. On the other hand, gammaglobulin levels, including IgA and IgM were low, and periodic replacement of gammaglobulin was necessary. At the age of 12, during the course of the treatment, she developed her first episode of otitis media, which developed into otitis externa. Lymphocyte surface antigen testing showed a significant reduction in memory B cells and a decreased response to immunization. With these observations, we suspected possible underlying CVID, and elected to perform genetic analysis, which revealed a missense mutation in NFKB1 that may have been associated with the etiology.

Conclusions: RTX therapy causes prolonged hypogammaglobulinemia associated with impaired B-cell differentiation in some patients exhibiting CVID-compatible clinical features. If an evaluation of genetic predisposition to immunodeficiency is performed prior to initiation of therapeutic intervention for refractory nephrotic syndrome, the choice of treatment options can be modified while considering any foreseeable risks.

Points of discussion: The necessity and usefulness of immunological screening and genetic analysis prior to RTX administration should be further explored, since it is difficult to distinguish hypogammaglobulinemia from CVID after RTX administration when the former is persistent.

Keywords: Refractory nephrotic syndrome, Hypoga- mmaglobulinemia, Rituximab, Common variable immunodeficiency

O-2

Persistent benign proteinuria associated with CUBN variants

Yun Young Choi[1], Yoon Sunwoo[1], HyunKyung Lee[2],
Jeesu Min[1,3], Yo Han Ahn[1,3], Hee Gyung Kang[1,3]

[1]Department of Pediatrics, Seoul National University Children's Hospital, Seoul, [2]Department of Pediatrics, Kangwon National University Hospital, Chuncheon, [3]Department of Pediatrics, Seoul National University College of Medicine, Seoul, Korea

Backgrounds: Causes of persistent proteinuria are diverse, usually indicating a disease of the urinary system. If left untreated, proteinuria may contribute to kidney damage through various mechanisms including oxidative stress and inflammation. Therefore, persistent proteinuria mandates investigation and intervention. In asymptomatic children, at first orthostatic proteinuria or tubular proteinuria is suspected. Upon excluding these, a kidney biopsy is considered to rule out glomerulopathy. However, sometimes histology turns out to be non-specific. The recent discovery of CUBN, encoding the membrane glycoprotein cubilin, sheds light on some of those cases. Since cubilin is a component of the cubilin-amnionless-megalin complex that is responsible for the receptor-mediated endocytosis of albumin in the proximal tubules, a defect of cubilin leads to a reduction in albumin reuptake, consequently results in albumin- dominant proteinuria. Interestingly, variants located at the N-terminal of CUBN result in severe proteinuria and megaloblastic anemia, whereas variants at the C-terminal are associated with benign, isolated proteinuria. Here we present five cases (M:F 3:2) with persistent proteinuria associated with homozygous or compound heterozygous C-terminal variants of CUBN.

Case: Children with proteinuria without nephrotic syndrome feature (edema, decreased serum albumin levels) were selected to get WES. Retrospectively, five CUBN mutation positive patients were included in this study. All patients presented with incidentally found isolated asymptomatic proteinuria, at their median age of 7 years (range 1.5 ~ 9). Their urine protein creatinine ratios were median 0.84 (0.57~2.03) mg/mg at presentation and did not change significantly over time regardless of RAS inhibition (median follow-up duration of 4 years [1 yrs~12yrs]). Their laboratory findings were also unremarkable at presentation or during follow-up for estimated GFR, serum albumin, lipid, hemoglobin, urine □2-microglobulin. None had hypertension, and kidney ultrasound showed normal kidneys. Among two patients, a kidney biopsy was done, which revealed no remarkable findings

Conclusions: These cases are similar to previously reported cases, indicating benign proteinuria associated with C-terminal variants of CUBN needs to be considered in such cases.

Points of discussion: 1. How does CUBN mutation site correlates with protein structure 2. Where CUBN protein localize 3. Which laboratory markers do we have to follow up with CUBN mutation patients 4. Asymptomatic proteinurea management

Keywords: CUBN, Proteinuria, Cubilin, Tubular proteinuria

O-3

A Novel MYH9 Mutation in a Chinese Child with Immune Complex-Mediated Glomerulonephritis and Literature Review

Jing Song[1,2,3,4], Qin Yang[1,2,3,4], Anshuo Wang[1,2,3,4], Sijie Yu[1,2,3,4], Qui Li[1,2,3,4], Mo Wang[1,2,3,4]

[1]Department of Nephrology, Children's Hospital of Chongqing Medical University, Chongqing, [2]Chongqing Key Laboratory of Pediatrics, Chongqing, [3]Ministry of Education Key Laboratory of Child Development and Disorders,National Clinical Research Center for Child Health and Disorders,China International Science and Technology Cooperation Base of Child Development and Critical Disorders,Chongqing, [4]Department of Rheumatology and Immunology, Children's Hospital of Chongqing Medical University, Chongqing, China

Backgrounds: MYH9-related diseases(MYH9-RD) are autosomal dominantly inherited disorders caused by mutations of the MYH9 gene encoding the non-muscle myosin heavy chain IIA (NMMHC-IIA). They are characterized by congenital thrombocytopenia, giant platelets and leucocyte inclusion bodies (Döhle-like bodies). Hearing impairment, cataracts, nephropathy and elevation of liver enzymes can also occur. In this article, we report an early-onset Chinese child with MYH9-associated immune complex-mediated glomerulonephritis and summarize renal involvement in the literature about MYH9.

Case: A Chinese boy had thrombocytopenia since birth, the hematological examination showed platelet fluctuation at 30-92×109/L, MPV of 8.5-12.7fL, giant platelets, and platelet level often decreases significantly during acute infection. He denied a family history of kidney disease, hematologic disorders or genetic metabolic disease and had no history of a bleeding tendency until his current admission. When he was 5 years old, he has diagnosed with nephrotic syndrome: hypoalbuminemia (serum albumin 25.9g/L), hyperlipidemia (TC 16.43 mmol/L, TG 3.85 mmol/L), massive proteinuria (urine protein "3+",24h urine protein quantitation 4026mg/ 24h). In addition, he had microscopic hematuria, hypertension (blood pressure 122/59mmHg),and CKD stage 2 (eGFR 77ml/ min/1.73m2). Ultrasonography demonstrated the echo of renal parenchyma was enhanced and the boundary of the cortex and medulla was not clear. The audiometric findings revealed moderate bilateral sensorineural deafness. The ophthalmological findings were normal. The renal biopsy was performed and renal pathology was shown in Figure 1. The results showed that the patient had immune-complex mediated glomerulonephritis with significant IgG(+++), IgA(+++), IgM(+), and C3 (+++) deposition in glomeruli and renal tubules. Whole-exome sequencing showed a novel c.2104C>G(Arg702 Gly) mutation in the exon17 of MYH9 gene. The p.(Arg702Gly) mutation substitutes an evolutionarily conserved amino acid in the protein's critical myosin globular head domain(Figure 2.A) and is predicted to be highly deleterious by Pymol,

PolyPhen2, and CADD(Figure 2.B). According to ACMG Standards, we estimated that sequence variant was pathogenic. One strong evidence (PS2), three moderate evidence (PM1, PM2, PM5), and two supporting evidence (PP2, PP3) met the ACMG criteria. There was little response to prednisone combined with captopril therapy during the first 4 weeks. Partial remission of urinary protein was achieved after 6 weeks of oral prednisone, captopril and tacrolimus. The patient was followed for 3 months, with abnormal 24h urinary protein quantitation (1375mg/ 24h), urine protein "3+", BUN 10.73mmol/L, Scr 99.00 μmol/L, UA 411.00 μmol/L, eGFR 63.45ml/min/1.73m2 (in stage-2 CKD) at the most recent follow-up visit. Extrarenal manifestations and clinical evolution of MYH9-RD are extremely variable: in some patients, mild to moderate thrombocytopenia remains the only manifestation of the disease throughout life, whereas other subjects develop over time complex syndromic pictures characterized by spontaneous bleedings, deafness and cataract. The literature review indicates that MYH9- related renal manifestations include proteinuria, hematuria, nephrotic syndrome, renal insufficiency. After the onset of the nephropathy, most patients will progress to CRF and ESRD rapidly. FSGS is the most common pathological type in children with MYH9-RD. The characteristics of renal pathology are mesangial proliferation, segmental glomerulosclerosis, inflammatory-cell infiltration and variable tubulointerstitial disease. And in the majority of cases, immunofluo- rescence is negative. Under electron microscopy include GBM abnormalities and fusion or effacement of podocyte foot processes.

Conclusions : We reported a novel pathogenic site c.2104C>G(Arg702Gly) mutation in the exon17 of MYH9 gene, accompanied by massive multiple immune complex deposited in the kidney which occurred at a young age, expanding the spectrum of MYH9 pathogenic variants.

Points of discussion: There are currently no effective interventions to slow the progression of MYH9-RD kidney injury to ESRD. In this case, we cannot explain the phenomenon of immune complex deposition in the kidney caused by MYH9 gene mutation, but its related pathogenic mechanism may indicate new therapeutic prospects.

Keywords: MYH9-related disorder; Immune Complex- Mediated Glomerulonephritis; Thrombocy-topenia; Gene mutation; Renal pathological; non-muscle myosin heavy chain-IIA

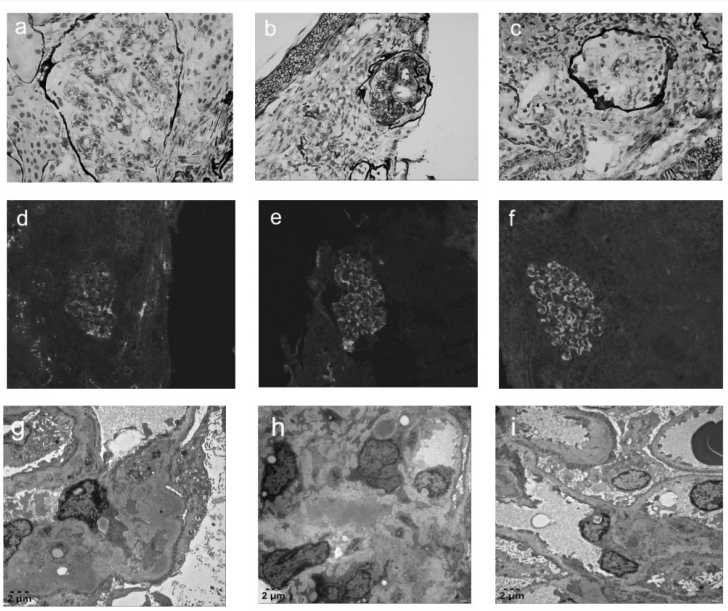
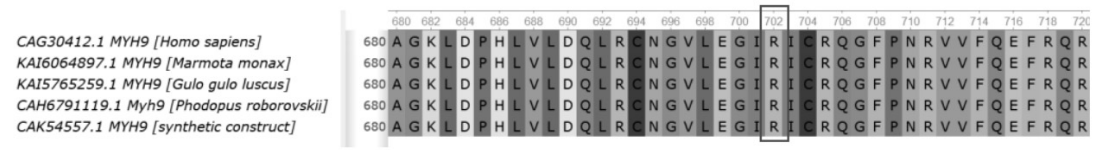


Figure 1. Kidney biopsy findings (a-c) Light microscopy revealed there was no obvious thickening of GBM, moderat proliferation in mesangial cells and mesangial matrix. A kidney biopsy specimen containing 2 glomeruli showe glomerulosclerosis and 1 glomerulus showed segmental glomerulosclerosis. Segmental endothelial cell hyperplasia wit open capillary loops. The renal tubulointerstitial lesions, including granular degeneration, vacuolar degeneration in rena tubules, and diffuse infiltration of inflammatory cells in the interstitial were also found. There was no obvious renal interstitia fibrosis. (d-f) Immunofluorescence examination indicated C3 (+++), IgA(+++), IgG(+++) deposits in glomeruli. (g-i) Massiv electron-dense deposition under mesangial cells and on some GBM and diffused fusion of podocyte foot processes wer observed on an electron microscope.



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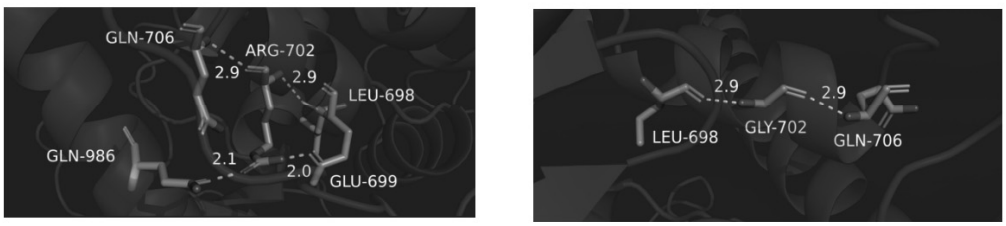

(a) Wild-type (b) Mutant

Figure 2.(A)Alignment of *Homo sapiens MYH9* and four other speies with the conserved residues boxed. (B)Tertiary structure prediction: After mutation, the number of hydrogen bonds changed and the spatial conformation changed significantly.

O-4

Multinucleated podocytes as a key clue to early diagnosis of cystinosis: Two case reports

Ayako Ogata[1], Saori Deki[2], Satomi Higashi[3], Toru Uchimura[4], Aya Inaba[4], Masako Otani[5], Shuichi Ito[6]

[1]Department of Pediatrics, Saiseikai Yokohamashi Nanbu Hospital, Kanagawa, [2]Department of Pediatrics, Fujisawa City Hospital, Kanagawa, [3]Department of Pediatrics, Saiseikai Yokohamashi Tobu Hospital, Kanagawa, [4]Department of Pediatrics and [5]Pathology, Yokohama City University Medical Center, Kanagawa, [6]Department of Pediatrics, Graduate School of Medicine, Yokohama City University, Kanagawa, Japan

Backgrounds: Cystinosis is a rare autosomal recessive lysosomal disease which causes various systemic disorders, particularly in the kidney and cornea. Oral cysteamine therapy prevents or at least delays the complications of the disease. However, the diagnosis of cystinosis, especially the juvenile nephropathic form, remains challenging because typical symptoms only become apparent in adulthood. We describe two cases of juvenile nephropathic cystinosis in which multinucleated podocytes served as a diagnostic clue to early diagnosis. We also studied the nephropathology of both cases to determine the characteristics of multinucleated podocytes.

Case: A 13-year-old girl was referred to our department because proteinuria was found on her school urine screening. Her medical history was unremarkable except for proteinuria at the age of 11 years, for which her parents did not consult her physician at that time. She was asymptomatic and had no visual impairment or hypertension. Urinalysis revealed proteinuria (urinary total protein/creatinine ratio: 0.6) and a high β-2 microglobulin level, but no other signs of Fanconi syndrome. She had no kidney insufficiency, with a serum creatinine level of 0.56 mg/dL and her estimated glomerular filtration rate was 108 mL/min/ 1.73 m2. Because her proteinuria had persisted for 6 months, a kidney biopsy was performed. Her biopsy specimen contained 34 glomeruli. Three glomeruli exhibited global sclerosis, and four showed segmental sclerosis. There was no crescent formation, and focal tubular dilatation and atrophy with interstitial fibrosis were partially observed. The most striking finding was the distinctive number of multinucleated podocytes, 15 of which were observed in all 34 glomeruli. The maximum number of nuclei in a podocyte was 10, and the mean number of nuclei per multinucleated podocyte was 4.4. Cystine crystals were observed on electron microscopy examination. Ophthalmological examination showed cystine crystals in her cornea. Her white blood cell cystine level was high, and she was diagnosed with juvenile nephropathic cystinosis. A genetic test is underway. She started oral cysteamine

treatment and showed almost no progression of the disease after 2 years. We studied another case of juvenile nephropathic cystinosis who had been diagnosed with Fanconi syndrome at the age of 3 years, but had not been evaluated or treated for several years. He underwent his first kidney biopsy at the age of 12 years. Twenty-five multinucleated podocytes were observed in 63 glomeruli. The maximum number of nuclei in a podocyte was 4, and the mean number of nuclei per multinucleated podocyte was 2.92. Electron microscopy revealed cystine crystals. Blood testing showed a high white blood cell cystine level, and ocular examination showed cystine crystals in the corneas. Mutations were found in CTNS gene. The patient was diagnosed with juvenile nephropathic cystinosis and started to take oral cytsteamine. He showed only mild kidney dysfunction (eGFR 75 ml/min per 1.73m^2) at the age of 20 years.

Conclusions: Because the prognosis of cystinosis depends on early treatment, it is important to recognize the unique pathological features of multinucleated podocytes as an essential key clue to the diagnosis of cystinosis.

Points of discussion: Characteristics of multinucleated podocytes, Approaches to early diagnosis of cystinosis

Keywords: multinucleated podocyte, juvenile nephropathic cystinosis, early diagnosis

O-5

Refractory hyperkalemia after kidney transplantation in a Korean pediatric patient

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Backgrounds: Renal tubular dysgenesis (RTD) is a rare and fatal disease caused by a genetic defect in the renin-angiotensin system (RAS) presenting poor or absent proximal tubule development. Mostly do not survive due to respiratory distress with anuria and severe hypotension, not responding to usual treatments. We report a Korean child with a non-fatal clinical course confirmed as RTD after kidney transplantation (KT).

Case: A 3-year-old Korean girl received deceased donor KT after two years of peritoneal dialysis with unknown etiology of renal insufficiency. She had a history of severe oligohydramnios at a gestational age of 17 weeks and was born at 32 weeks of gestation by emergency cesarean section due to persistent oligohydramnios. A week after birth, she underwent continuous renal replacement therapy for 7 days due to diuretic-resistant oliguria. Even though diuresis recovered, she reached end-stage renal disease at the age of seven months. Immediately after KT, she had no complications and was discharged. A week after discharge, she presented with vomiting and poor oral intake with polydipsia and polyuria. Severe hyponatremia, hyperkalemia, azotemia, and metabolic acidosis were detected. These findings were relieved after supportive care with normal saline hydration, kalimate, and sodium bicarbonate medication. Similar events repeated twice more. With suspicion of polyuria caused by tubulopathy of the native kidney, both native kidney nephrectomies were done at 80 days after KT. Even after nephrectomy, similar events persisted. To verify the cause of recurrent and uncontrolled hyperkalemia and metabolic acidosis, renin activity and aldosterone level were evaluated. As a result, high renin activity (25.3ng/ml/hr) and low aldosterone level (2.6ng/dL) were detected. For further management, fludrocortisone was started, and electrolyte imbalance and metabolic acidosis gradually improved. For the hypoaldosteronism etiology evaluation, whole- exome sequencing was done, and homozygous ACE gene mutation (p.Ser486Phefs Ter29) was detected and diagnosed with RTD.

Conclusions: Even though hyperkalemia is a common complication in KT patients, if refractory hyper-

kalemia persists in post-KT patients, RAS evaluation and the genetic study is necessary for the suspicion of RTD.

Points of discussion: 1. When to suspect RTD and undergo a genetic test in this patient? 2. Indication of a genetic test in pediatric chronic kidney disease with unknown etiology planning to undergo KT.

Keywords: Kidney transplantation (KT), Hyperkale- mia, Renin-angiotensin system (RAS), Renal tubular dysgenesis (RTD)

O-6

An 8-month-old boy with infantile nephrotic syndrome caused by semaphorin 3B-associated membranous nephropathy

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Backgrounds: Novel target antigens for primary membranous nephropathy (MN), such as PLA2R, THSD7A, Nell-1, and EXT1/EXT2 have been discovered in the last few years. Semaphorin 3B (sema3B) was reported to be a new target antigen in 2020. The limited number of reported cases of sema3B-associated MN hinders the clinical characteristics, prognosis, and treatment strategies.

Case: The patient was an 8-month-old male infant who presented with severe proteinuria and hypertension. Genetic screening for podocyte-related genes was negative. He was treated with prednisolone (PSL) for nephrotic syndrome; however, remission was not achieved within 4 weeks. He was diagnosed with steroid-resistant nephrotic syndrome (SRNS) and underwent kidney biopsy. Pathological examination revealed MN with IgG deposits on both the glomerular basement membrane (GBM) and the tubular basement membrane (TBM). He was treated with CsA in addition to PSL and achieved complete remission. However, frequent relapses occurred after the discontinuation or tapering of immunosuppressants. Two years after treatment initiation, a second biopsy was performed and showed a worsening of the disease. In addition, IgG4 subclass staining was negative, but IgG1 was positive. He required treatment with several immunosuppressants to achieve complete remission. To uncover the pathophysiology, we performed additional immunostaining for sema3B with the clue of IgG deposit on the TBM that is reported to be the pathological character in sema3B- associated MN. As a result, we confirmed the diagnosis of sema3B-associated MN.

Conclusions: Sema3B-associated MN should be included in the differential diagnosis of young children with SRNS, and our report was the first case of infantile nephrotic syndrome (INS). Sema3B-associated MN may be needed strong treatment in some case.

Points of discussion: Despite the rarity of childhood MN, in sema3B-associated MN, 8/12 of cases

were in children. Our patient is the youngest case and the first case manifesting INS. In a report for primary MN in children, 67% were on immunosuppressants and 5% developed ESKD. On the other hand, in sema3B-associated MN, 82% were on immunosuppressants and 18% developed ESKD. Our patient had been treated with PSL, MPT, CsA, MZR, and RTX; however, he showed a refractory immunosuppressants-dependent course.

Keywords: Membranous nephropathy, Semaphorin 3B, Pediatric

O-7

Single-cell transcriptomics reveal a B cells-mediated immune mechanism of the onset of immunoglobulin-A vasculitis nephritis: gut-homing B cells migrate to the kidney?

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Backgrounds: Immunoglobulin-A vasculitis (IgAV) is an inflammatory small-sized blood vessel disease, more common in children. The classic clinical triad of IgAV consists of purpura, arthralgias and gastrointestinal tract involvement. Nephritis is also common in affected patients. Perivascular and mesangial predominant deposition of Gd-IgA1 immune complexes are the defining pathophysiologic feature of IgAV nephritis, supporting the hypothesis that this nephritis is predominantly a B cell-involved disease. Nonetheless, it remains unknown how kidney resident immune cells participate in the pathogenesis of nephritis.

Case: Here, we perform single-cell RNA sequencing (scRNA-seq) on kidneys collected from a patient with aggressive IgAV nephritis and normal sample. The patient, a 6-year-old girl, presented with abdominal pain, arthralgia, purpuric rashes on both lower limbs, hematuria (urine sediment of red blood cells >100/ HPF), severe proteinuria (8.0 g/24h), hypoproteinemia (27.8 g/L) and edema with a serum creatinine of 32 μmol/L. The results of tests for ANCA and cryoglobulins were negative, all other autoantibodies were negative, complement was within normal range, and serum IgA was within the reference range. She had no history of urinary abnormalities or purpura in the past. A kidney biopsy was performed on the second day of admission, after which methylprednisolone 500 mg was administered intravenously for 3 days, followed by daily prednisolone 40 mg and monthly intravenous cyclophosphamide. After the initiation of the steroid, the patient's purpura, arthritis, and abdominal pain improved, but hematuria and proteinuria remained. In the renal tissue specimens, all 19 glomeruli showed mesangial and endocapillary proliferative changes, 7 of which were accompanied by cellular crescent for- mation. Focal tubulo-interstitial lesions were detected, including tubular atrophy, interstitial fibrosis, and a large number of mononuclear cells infiltration. Imm- unofluorescent staining on a frozen section showed IgA and C3 dominant deposition in the mesangial areas. An electron microscopy examination revealed cell proliferative changes with an electrondense deposit in the mesangial area

and extensive foot process effacement of podocytes. The results of scRNA- seq showed that the number of both classical monocytes and lymphocytes significantly increased in IgAVN patient. Interestingly, the number of T and NK cells decreased, while B cells increased significantly in IgAVN patient. A distinctive B cell subset was detected in IgAVN patient, which had high levels of CXCR4. Higher constitutive levels of CXCL12 (the ligand of CXCR4) in the intestinal Peyer's patches than in other lymphoid or- gans. Moreover, the DEGs of B cells were enriched in different viral infection pathways.

Conclusions: We dissected the immune landscape of pediatric IgAVN at the single-cell resolution, which provides new insights into pathogenesis and developing new immunotherapy against IgAVN.

Points of discussion: 1. Do gut-homing B cells abnormally migrate to the kidney at the onset of IgAV nephritis? Is the number of CXCR4high B cells positively associated with the clinic-pathological parameters, including the levels of urine protein creatinine ratio, the levels of IgA deposition in the mesangial areas, and the number of crescents? 2. Is IgA vasculitis associated with viral infection such as COVID-19? 3. Is targeting B cell therapy feasible? Is rituximab effective for induction and maintenance of long-lasting remission in severe IgAV with aggressive renal involvement?

Keywords: immunoglobulin-A vasculitis nephritis, single-cell RNA sequencing, CXCR4

O-8

Renal complications of Pediatric Glycogen Storage Disease, type I

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Backgrounds: Glycogen storage disease (GSD) type I is an inherited disorder in which glucose-6-phosphatase enzyme complex is deficient. Glucose-6-phosphatase is predominantly present in the liver, proximal renal tubule and intestine, so several renal complications have been reported in type I GSD patients. Enlargement of the kidney is the earliest finding detected due to the accumulation of glycogen in the kidneys. Nephrolithiasis, hyperuricemia, hyperlipidemia and proximal tubular dysfunction have also been described in type I GSD patients. Although impaired renal function is also one of major complications in type I GSD patients, the etiology is not well known yet.

Case: A 15-year-old girl visited our hospital with proteinuria. She was diagnosed with type I glycogen storage disease around the age of 1 year and had regular outpatient visits at another hospital. She grew normally through optimal treatment including dietary therapy, but she has neglected dietary therapy in recent years. She had proteinuria from 4 years ago and took ACEi and ARB (stopped taking the drug 2 months ago due to increased BUN and hypercalcemia). She did not have hypertension and clinical symptoms related to proteinuria. The laboratory test results at the first visit were as follows; WBC 3,570/uL, Plt 450K/uL, BUN 9.8 mg/dl, serum creatinine 0.33 mg/dl (eGFR 200 mL/min/1.73m2), sodium 135 mmol/L, potassium 4.2 mmol/L, total protein 8.2 g/dL, albumin 5.0 g/dL, calcium 10.2 mg/dL, phosphorus 4.9 mg/dL, AST/ALT 47/67, random glucose 78 mg/dL, total cholesterol 205 mg/dL, TG 326 mg/dl, LDL-chol. 128 mg/dL, uric acid 8.7 mg/dL, VBGA pH 7.40- PCO2 38.6-HCO3- 24.0, spot urine P/Cr ratio 1.3 g/g Cr, spot urine beta- 2-microglobulin/Cr 12.0 mcg/g and spot urine Ca/Cr 0.17 g/g Cr. Abdominal ultrasonography showed hepatosplenomegaly and marked enlargement of both kidney with increased echogenicity (RK: 16.2 Cm, LK: 15.1 Cm). The renal biopsy results showed focal deposition of glycogen particles in proximal tubular epitheliums, glomerular enlargement and mild interstitial fibrosis. After the biopsy, she started taking ARB and proteinuria is gradually improving.

Points of discussion: 1. What is the pathogenic mechanism of proteinuria in this patient? 2. What are the prevention and treatment of renal complications in GSD?

Keywords: Glycogen Storage Disease, Renal complication

O-9

A Case of Thrombotic Microangiopathy Caused by Hypereosinophilic Syndrome

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Backgrounds: Hypereosinophilic syndrome (HES) are a group of disorders marked by the sustained hypereosinophilia and can cause damage to multiple organs. Reports of kidney involvement are rare, especially in infant. We reported a case of infancy onset renal thrombotic microangiopathy (TMA) caused by hypereosinophilic syndrome.

Case: A one-year-old girl, was admitted to hospital with "intermittent tachypnea and edema for 10 months, hepatomegaly and oliguria for 2 months". The girl suffered with heart failure after gastrointestinal infection when she was 3-month-old, and then worsened repeatedly after infection. Two months before admission, edema, hepatomegaly, oliguria, renal dysfunction, anemia, and thrombocytopenia were observed, and peritoneal dialysis was started. After admission, multiple organs dysfunction was detected, including heart, kidney, respiratory system, digestive system, blood system, skin, etc. It was found that peripheral blood eosinophilia was sustained increased (>1.5*109/L), and the proportion of bone marrow eosinophils was significantly increased, which was consistent with the diagnosis of hypereosinophilia. Renal biopsy showed TMA. A small amount of eosinophil infiltration was observed in glomeruli and interstitial. Eosinophil infiltration was also observed in the interstitial of esophagus, antrum, and duodenum. Meanwhile, multiple plaques in the subcutaneous were found, which was considered eosinophilic panniculitis. HES was diagnosed because of hypereosinophilia and target organs damage. The etiological examination of HES was completed, but no secondary or primary etiology was found. Predniso- lone was administered orally at 1mg/kg. After treatment, eosinophil decreased significantly, and renal function recovered. The peritoneal dialysis tube was removed 2 months ago, and the patient is now followed-up.

Conclusions: This is the youngest HES child with kidney involvement until now. The pathological mechanism may be that eosinophil toxic granulocyte products damaged vascular endothelial cells and then led to TMA. Etiological examination can identify the cause of HES and guide target treatment.

Early diagnosis and treatment can improve prognosis.

Points of discussion: Etiology and pathogenesis of TMA caused by HES, future treatment for this girl.

Keywords: Hypereosinophilic syndrome (HES), th- rombotic microangiopathy (TMA), peritoneal dialysis

Poster Presentation

P1-1

ANCA-associated Vasculitis Followed by Alveolar Proteinosis in a 9-Year-Old Boy

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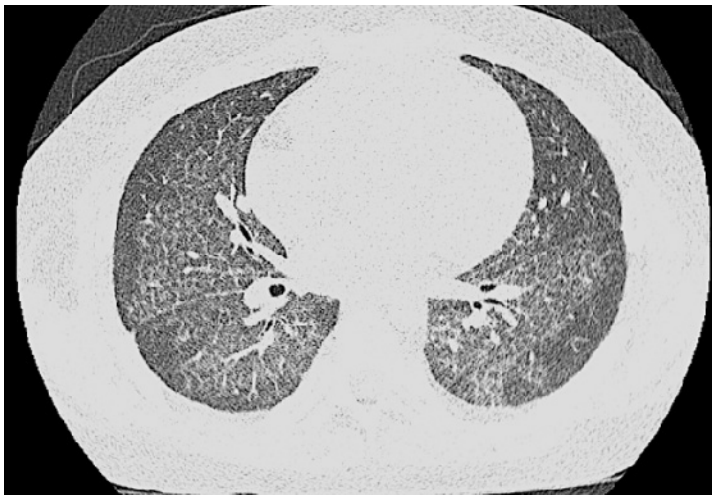
Backgrounds: Both pulmonary alveolar proteinosis (PAP) and antineutrophil cytoplasmic antibody (ANCA)- associated vasculitis (AAV) are rare diseases and can lead to hypoxemia. We describe a case of these two rare pathologies occurring together.

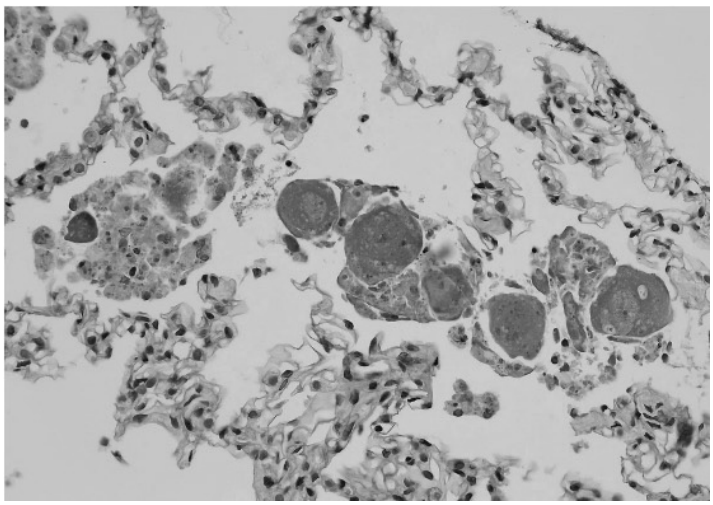
Case: A 9-year-old boy had received a diagnosis of microscopic polyangiitis (MPA) 7 months previously, confirmed by kidney biopsy and an elevated serum ANCA level. His initial presentation of MPA included renal failure and alveolar hemorrhage with symptoms of cough, hemoptysis, and lethargy. He was treated with high-dose methylprednisolone, cyclophospha- mide combined with rituximab. His respiratory syndrome and hypoxemia disappeared one month later. But four months ago, routine ECG monitoring showed that he had hypoxemia after falling asleep. Despite rituximab and azathioprine was administered for maintenance of MPA remission, as well as using empirical antimicrobial therapy for infection, his condition failed to improve. At the time of our evaluation, his ANCA profile and the urinary sediment results were normal. A CT scan of the chest revealed bilateral, diffuse geographic ground- glass opacities (GGO) with crazy-paving appearance. A cryoprobe transbronchial lung biopsy revealed alveolar accumulation of proteinaceous material that was positive using periodic acid-Schiff stain without active vasculitis. We stopped the maintenance therapy of MPA and initiated the use of recombinant granulocyte-macrophage colony stimulating factor (GM-CSF) by inhalation. At a 5-month follow-up visit, the patient was asymptomatic with obvious resolution of the changes on the chest radiograph.

Conclusions: To our knowledge, this is the first reported case of MPA and PAP occurring in pediatric patients simultaneously. We suggest that the alveolar macrophage become dysfunctional in surfactant clearance due to immunosuppressive treatment. For ANCA vasculitis patients with hypoxemia, early diagnosis and treatment may improve their prognosis.

Points of discussion: The possible pathogenesis and best treatment for this child

Keywords: pulmonary alveolar proteinosis, antineutrophil cytoplasmic antibody-associated vasculitis, hypoxemia, immunosuppressive treatment





P1-2

A case of membranous + mesangial proliferation nephritis showing full-house pattern on immunofluorescence with good clinical course

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Backgrounds: Membranous nephritis is a disease caused by deposition of immune complexes under the glomerular basement membrane and complement activation. It is classified into primary and secondary group, and SLE belongs to secondary group. In the case of lupus nephritis, immunofluorescence (IF) often shows "full- house pattern", which is the variegated appearance of the deposits such as IgG, IgA, C3, C4, C1q. We report a case of "membranous + mesangial proliferation with full-house pattern nephritis", which hasn't turned out to be SLE for two years.

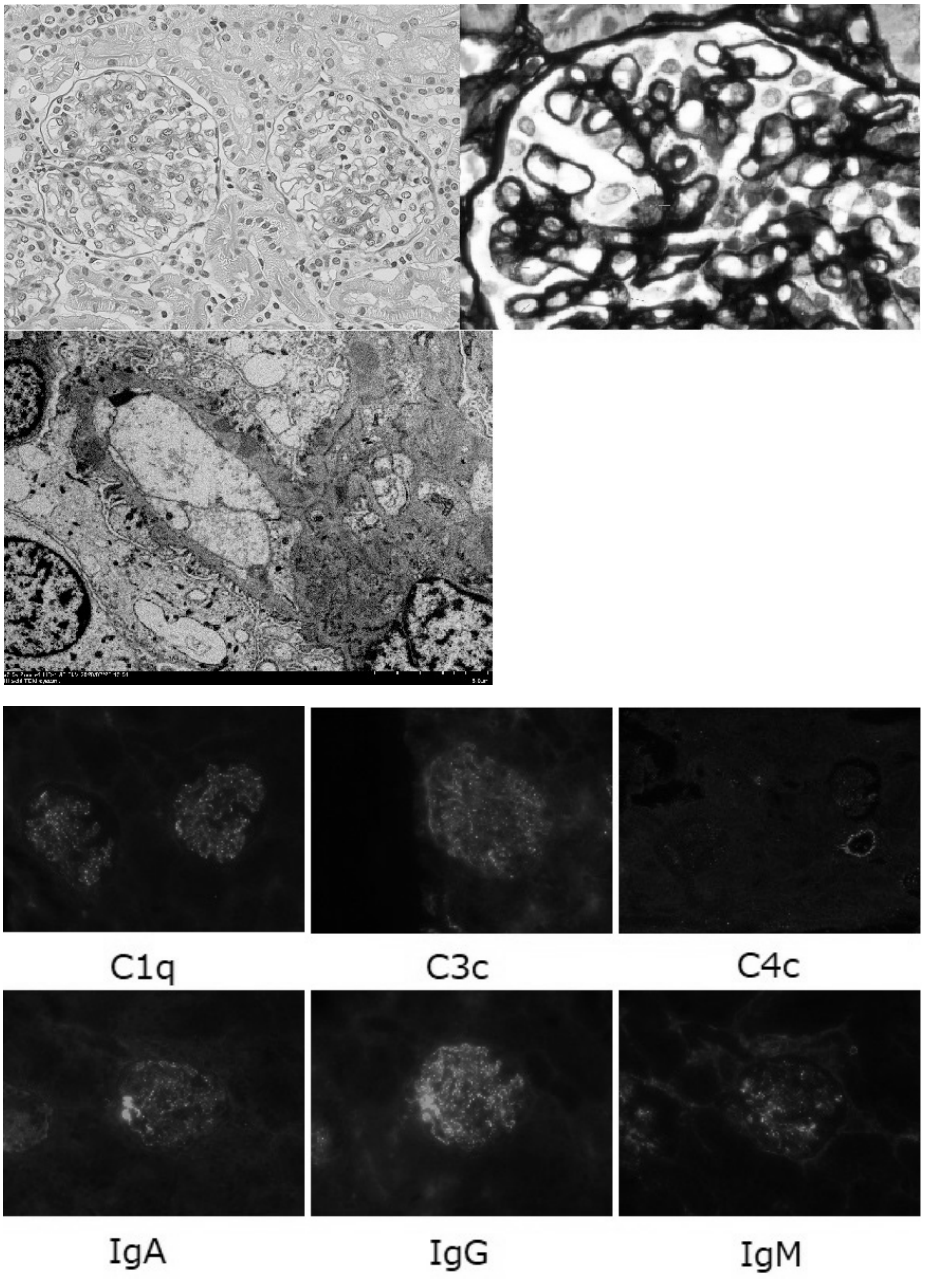
Case: The patient was a 3-year-old girl with hematuria (RBC>100/HPF), proteinuria(UP/Cre : 0.65-0.75 g/gCr) and RBC cast(+). Serum albumin and IgG decreased down to 3.1 g/dL and 315 mg/dL, respectively. Total cholesterol elevated 292 mg/dL. There were neither hypocomplementemia nor positive antibodies related to SLE. Renal pathological diagnosis was "Diffuse global/segmental mesangial proliferation, mild to moderate + diffuse segmental/global bubbling + immature glomeruli (4/43)". It was atypical as a membranous nephritis because it also showed mesangial proliferation and IF showed full-house pattern. Electron microscope showed Electron-dense deposit (EDD) varied in size and EDD was also observed in mesangial cells. The wire-loop lesion was not observed. Therefore, secondary membranous nephritis was more likely suspected than primary one. The diagnosis was considered to be "mainly membranous glomerulonephritis", while in the case of lupus nephritis, the type was classified as ClassV+II. Since then, the patient has been treated with lisinopril alone, and there were improvements in blood albumin, IgG, and Total-cholesterol, as well as improvements in proteinuria and hematuria (RBC=5-9/HPF). Over the next two years, ANA and anti-DNA antibodies have remained within the normal range, and no other findings related to SLE have occurred.

Conclusions: In this study, we experienced a case with a good clinical course of membranous + mesangial proliferation nephritis showing "full-house pattern". Even if "full-house pattern" was observed in a case, continuous observation regarding other autoantibodies and physical findings must also be es-

sential for the differentiation from SLE. The fact that her clinical findings improved only by lisinopril seems to be compatible with a course of membranous nephritis, although it is still necessary to pay attention to the possibility that it is secondary in the future.

Points of discussion: 1. Can this case be considered as "membranous + mesangial proliferation nephritis" rather than SLE? 2. Please give us some advices regarding additional special examinations including antibodies for the final diagnosis in this case.

Keywords: membranous nephritis, glomerulonephritis, full-house pattern, lisinopril



P1-3

Passenger lymphocyte syndrome in a minor ABO-Incompatible Pediatric Kidney Transplant Recipient Receiving Plasmapheresis: A case report

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Backgrounds: People with type O blood are considered as universal organ donors compatible with any other blood types. However, in the case of minor ABO-incompatible transplantation, immune-mediated hemolysis can occur due to concomitant transfer of donor B- lymphocytes together with the allograft. These so-called passenger lymphocytes may produce antibodies against the recipient's red blood cells, causing hemolytic anemia, known as the passenger lymphocyte syndrome (PLS). Herein, we report a case of gastrointestinal involvement of PLS following minor ABO-incompatible kidney.

Case: A 6-year-old boy (blood type A+) underwent a kidney transplant from his father (O+). His kidney transplant surgery went well with good primary graft function. Immunosuppression was composed of prednisolone, tacrolimus, and mycophenolate mofetil after induction with basiliximab. On day 6, the patient developed a fever with no explainable causes and persisted despite broad-spectrum antibiotics. On day 11, he presented abdominal pain, hematochezia, and severe diarrhea, with sudden Hb drop from 9.3 g/dℓ to 5.2 g/dℓ, increased LDH, decreased haptoglobin, and mildly elevated total bilirubin. An abdominal CT scan revealed no signs of active bleeding but segmental wall thickening in the rectosigmoid colon, suggesting colitis. He was managed supportively. On day 13, platelet count abruptly fell to 58×103/㎕ along with C4 (3mg/dℓ). Evidence of thrombotic microangiopathy could not be found, with no compatible lesion on kidney biopsy and no schistocytes on peripheral blood smear. On day 20, direct antiglobulin test was positive to IgG, anti-A IgG/M titer was 32/2, and an anti-A antibody elution test was strongly positive (3+), consistent with PLS. To remove the anti-A antibodies, the patient received five times plasmapheresis, resulting in disappearance of anti-A antibody and clinical recovery on day 40.

Conclusions: Although rare, PLS should be part of the differential diagnosis when evaluating post-transplant immune-mediated hemolysis. Treatment of PLS is mainly supportive; however, plasmapheresis can be considered.

Points of discussion: How can we predict and prevent PLS Is PLS a risk of poor outcome?

Keywords: Passenger lymphocyte syndrome, immune-mediated hemolysis, pediatric kidney transplantation, plasmapheresis.

P1-4

A case of congenital syphilis with infantile nephrotic syndrome as the first manifestation

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Backgrounds: Recently, the number of cases of syphilis is increasing again all over the world, including Japan. Congenital syphilis (CS) occurs due to transplacental transmission of Treponema pallidum, and is recognized as one of TORCH infections. Early CS generally manifests during the first 3 months of life. Common symptoms include characteristic vesiculobullous eruptions, lymphadenopathy, hepatosplenomegaly, failure to thrive and blood-stained nasal discharge; renal disorder is rare. Here, we report a boy with CS who presented with infantile nephrotic syndrome (INS) as the first manifestation, who was not diagnosed until his mother's next pregnancy.

Case: Our patient was born at term with an uncomplicated perinatal course. His mother's serological syphilis test was negative at the beginning of her pregnancy. At the age of 4 months, he was admitted to a previous hospital because of his fever. His blood examination showed C-reactive protein (CRP) of 4.7mg/dL, serum albumin of 2.4g/dL, and serum creatinine of 0.22 mg/dL. His urinalysis showed proteinuria of 10.3g/g creatinine and erythrocytes of 50-99 per high power field. Five days later, he was transferred to our hospital because he was diagnosed as having infantile nephrotic syndrome (INS). No generalized edema nor failure to thrive were observed. Physical findings showed no cutaneous symptoms, lymphadenopathy nor hepatosplenomegaly. A few days later, his fever and urinary abnormality were completely improved without treatment. A renal biopsy was not performed because of the remission of INS. He had no genetic mutations in targeted gene panel analysis with INS. During the follow-up period, his CRP was re-elevated to 11.6mg/dL, but without his showing any symptoms including fever or proteinuria. His blood and urine cultures were negative. He showed only nonspecific inflammation in a contrast-enhanced computed tomography (CT) scan of the thorax and abdomen, brain magnetic resonance imaging examination, bone marrow examination, positron emission tomography-CT scan and cytokine profiles. Although rheumatoid factor and anti-cardiolipin antibody were positive temporarily, the elevated CRP level improved without treatment. He had neither poor weight gain nor psychomotor retardation. At the age of 21 months, his mother became pregnant with the sec-

ond child and was found positive in a serological syphilis test at a gynecological checkup. A re-interview of his parents' medical history during pregnancy of our patient revealed that his mother had a systemic skin eruption and his father developed a pudendal ulcer. The rapid plasma reagin test and treponema pallidum hemagglutination assay (TPHA) in his serum were positive at 260 R.U. and 21,810 T.U., respectively. Alth- ough TPHA in his cerebrospinal fluid was positive at 12.6 T.U., the later PCR analysis was negative. We diagnosed him with CS and treated with penicillin G for 10 days. Now, he is 27 months old and is developing normally.

Conclusions: CS is a multisystem infection including syphilitic nephropathy. When a patient with INS is identified, transplacental infections such as TORCH infections should be considered along with genetic predisposition. Because CS cannot be ruled out even if there is a negative maternal serological syphilis test in early gestation, it is necessary to become familiar with the symptoms and examinations of CS.

Points of discussion: His INS, developing as one symptom of CS, went into spontaneous remission. It is not clear that steroid therapy is recommended for secondary nephrotic syndrome due to syphilis.

Keywords: infantile nephrotic syndrome, Congenital syphilis

P1-5

Age is not a risk factor for persistent hypogammaglobulinemia with rituximab treatment in young children

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Backgrounds: Rituximab (RTX) is relatively safe and effective to maintain remission of childhood-onset nephrotic syndrome. However, prolonged hypogammaglobulinemia has been reported as an adverse event. Stratified analysis of age has not been conducted, although age and serum IgG level at the time of initial administration are considered risk factors. Therefore, we attempted a stratified analysis of risk factors for prolonged hypogammaglobulinemia.

Case : From September 2014 to August 2022, 32 patients (male: 21, female: 11, median age at RTX initiation: 13.1 years) were treated with RTX at our hospital. Of the 32 patients, 11 (32.3%) had persistent hypogammaglobulinemia (IgG <600 mg/dL for more than 6 months). Immunoglobulin supplementation was performed in 4 patients (11.7%), 2 of whom received subcutaneous immunoglobulin. Even after RTX discontinuation, seven patients (21.9%) tended to have lower IgG, five of these seven patients who received repeated RTX. Patients in the prolonged group had a lower age at treatment (10.8 vs. 14.6 years, p=0.054) without significance, and a significantly lower serum IgG level at the initial dose (418 vs. 642 mg/dL, p=0.037) compared to the non-prolonged group. The patients older than 16 years (6 patients) had not persistent hypogammaglobulinemia. In the patients younger than 16 years, there is no difference in the age at RTX initiation between the prolonged and non-prolonged groups (10.8 vs. 10.8 years, p = 0.42), the prolonged group had significantly lower serum IgG level at the initial dose (418 vs. 632 mg/dL, p=0.046). There was no difference in the number of patients who received repeated doses or the total number of doses given between the prolonged and non-prolonged groups.

Conclusions: Prolonged hypogammaglobulinemia was not seen in patients aged 16 years or older, so we performed a stratified analysis by dividing them into two groups: those aged 16 years or older and those aged <16 years. In that analysis, age was not a risk factor in the <16-year-old group, but serum IgG was a risk factor at initial administration.

Points of discussion: RTX may be safely administe- red in younger children, and the risks should be reconsidered in younger children.

Keywords: childhood-onset nephrotic syndrome, Ri- tuximab, persistent hypogammaglobulinemia

P1-6

A case of lupus nephritis in a 4-year-old female with disproportionate chromosomal abnormalities

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Backgrounds: Systemic lupus erythematosus (SLE) is a systemic inflammatory disorder with an autoimmune etiology, in which both genetic and environmental factors are involved. Genome-wide association studies have unraveled over 100 genetic loci associated with the risk of SLE.

Case: The patient was a four-year-old female with unbalanced chromosomal abnormalities including a partial trisomy of the short arm of chromosome 10 and partial monosomy of the long arm of chromosome 20 [46, XX, der(20)t(10;20)(q24;p13)]. She was diagnosed with aTTP at the age of 2 years and received treatment with prednisolone (PSL), intravenous immunoglobulin, fresh frozen plasma (FFP), and ritu- ximab. She later developed ITP and was treated with PSL and FFP at the age of 3 years. At the age of 4 years, she had a fever that lasted for two weeks. Blood tests were performed, and the results showed a WBC count of 2,600/μL, antinuclear antibodies of 1:160, and double-stranded-DNA antibodies of 710 IU/ml. She had a nephrotic syndrome with a urinary protein-to-creatinine ratio of 15.58 g/gCr, serum albumin of 1.4 g/dL, and no hematuria.

She satisfied 4 out of 11 items of the American College of Rheumatology 1997 revised classification criteria for SLE, although the extrarenal lesions were only faint discoid erythema. Intravenous pulse methylprednisolone was initiated because the liver dysfunction and high ferritin levels suggested that the condition was similar to macrophage activation syndrome. A renal biopsy was performed at the end of the second course of the pulse therapy. Light microscopy revealed mild to moderate mesangial hypercellularity and partial endocapillary proliferation without sclerotic glomeruli or crescentic formation. Immunoflu-orescent assay showed significant staining of IgG, C3, and C1q in the capillary and mesangial lesions of the glomeruli. Electron microscopy demonstrated numerous subepithelial and intramembrane deposits, which were predominantly mesangial and subendothelial immune deposits. Foot process loss and virus-like particles were also observed. Based on these findings, she was diagnosed with lupus nephritis type IIIA+V. Myco-phenolate mofetil was started and then cyclosporine was

added to the treatment regimen, followed by three courses of pulse therapy, but severe proteinuria occurred.

Conclusions: The incidence of SLE and lupus nephritis presented in this case is considered to be rare in terms of the presence of chromosomal abnormalities, the history of aTTP and ITP, lack of extrarenal lesions, and absence of hematuria. The genetic background could influence these pathologies.

Points of discussion: We want to discuss the future treatment strategies and the relationship between genetic abnormalities and the onset of SLE.

Keywords: SLE, lupus nephritis, aTTP, ITP

P2-1

A child with crescentic glomerulonephritis following the Pfizer-BioNTech COVID-19 Vaccine

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Backgrounds: There are few reports about renal complications after the COVID-19 vaccine. We report a pediatric case who were diagnosed as crescentic glomerulonephritis after the Pfizer-BioNTech COVID-19 Vaccine

Case: A sixteen-year girl was admitted due to dyspnea and headache. She had received a second Pfizer- BioNTech COVID-19 Vaccine about a month ago. She had experienced fever, nausea, vomiting, and dyspnea on exertion after the vaccination, which persisted for a week. A right temporal headache had developed after two weeks. She was transferred to our hospital because blood tests revealed severe azotemia and increased cardiac enzymes in a local hospital. Her blood pressure was 155/89 mmHg on admission. Her weight increased by 7 kg in a month. Laboratory findings were as follows. BUN/Cr 9.57/66 mg/dL, CK/LD 410/320 IU/L, BNP 1167 pg/mL. She also had hematuria and proteinuria (urine protein/Cr 9.1). Electro- cardiography showed sinus tachycardia. The result of echocardiography was grade I mitral regurgitation with normal cardiac function. Renal doppler revealed swelling and increased echogenicity of both kidneys with increased resistive index. Cardiac MR results were early minimal fibrosis of previous myocarditis. We started hemodialysis. A kidney biopsy was done, and the results were diffuse extracapillary proliferative glomerulonephritis with diffuse crescent formation. We treated her with methylprednisolone pulse therapy with subsequent oral steroids, mycophenolate mofetil, and angiotensin-converting enzyme inhibitor.

Conclusions: We report a first pediatric case of rapidly progressive glomerulonephritis, which developed after the Pfizer-BioNTech COVID-19 Vaccine.

Keywords: COVID-19 Vaccine, crescentic glomerulonephritis, children

P2-2

Two cases of hypertensive crisis in adolescents following mRNA COVID-19 vaccination

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Backgrounds: In response to the global COVID-19 pandemic, vaccines were developed and approved at a record speed. However, numerous cardiovascular adverse events have been reported. We present two adolescent cases who developed a hypertensive crisis following NT162b2 mRNA COVID-19 vaccination.

Case: Patient 1 was an 18-year-old male who complained with elevated blood pressure that was discovered a day after 2nd NT162b2 mRNA COVID-19 vaccine. His blood pressure was 190/125 mmHg. He denied the relevant past medical history. He remembered his systolic blood pressure as 130 mmHg a year ago. His father had hypertension. He was obese (BMI 29.2 kg/m2). Renal panel, renin, aldosterone, thyroid function test, and metanephrines were all within normal ranges. The level of AST and ALT was 48 IU/L and 80 IU/L, respectively. The urine test was negative. Kidney sonography was unremarkable. A labetalol continuous infusion was started and his blood pressure was decreased gradually. It was switched to losartan. Echocardiography showed no left ventricular hypertrophy. There were no abnormalities on the fundoscopy. After 5 months, his blood pressure was maintained at 135/81 mmHg with 100 mg daily losartan. Patient 2 was an 18-year-old male who complained with palpitation after 1st NT162b2 mRNA COVID-19 vaccine. Initial blood pressure was 178/109 mmHg, and his heart rate was 75 beats/min. He had a family history of autosomal dominant polycystic kidney disease in his mother and grandmother. He knew his enlarged kidneys, however, he did not follow up on it. Blood test including hormones and urine test were all normal. Kidney sonography showed diffusely increased size kidneys with multiple variable sized innumerous cysts, and it was compatible with autosomal dominant polycystic kidney disease. A labetalol continuous infusion was started and it was switched to enalapril. No end organ damage due to high blood pressure was observed on the echocardiography and fundoscopy. Blood pressure was 137/85 mmHg with 100 mg daily losartan in the outpatient clinic after 5 months. He received 2nd vaccination and his blood pressure did not rise.

Conclusions: It is warranted to measure blood pressure in adolescents at high risk of hypertension after mRNA COVID-19 vaccination.

Points of discussion: Did mRNA COVID-19 vaccination cause hypertensive crisis in these cases?

Keywords: COVID-19, Vaccines, hypertension

P2-3

A case of nephronophthisis caused by INVS gene mutation in a 6-year-old with non-dialysis-dependent status

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Backgrounds: Infantile nephronophthisis (NPHP type 2) is caused by mutations in the inversin (INVS) gene, often leads to end-stage renal disease (ESRD) by the age of 5 years, and is characterized by renal enlargement. We report a case of a 6-year-old with a non-dialysis-dependent status despite the presence of a homozygous INVS gene mutation.

Case: We report a case of a 6-year-old girl who presented with pyuria at her 3-year medical checkup. She was noted to have metabolic acidosis, renal dysfunction (Cr-eGFR 65 ml/min/1.73m^2), and severe tubular proteinuria. Ultrasonography showed bilateral renal enlargement and hyperechogenicity in the medulla. When she was 4 years and 4 months old, we performed genetic screening using a tubular disease-related gene panel, but no gene mutations were detected. At 5 years old, the patient underwent a renal biopsy that revealed the pathological findings of NPHP. When she was 6 years old, we performed genetic screening for congenital anomalies of the kidney and urinary tract with targeted sequencing using a next-generation sequencer, and a cystic kidney disease-related gene panel detected an INVS mutation. Sanger sequencing was used to confirm the mutation with an automated DNA sequencer. The genetic analysis revealed that the patient had a homozygous nonsense mutation c.2695C>T, p.Arg899Ter in exon 14 of the INVS gene. At the age of 6 years and 9 months, she had a renal function of Cr-eGFR 38.93 ml/min/ 1.73m^2.

Conclusions: If the clinical subtype and phenotype do not match, the possibility of a different subtype should be considered, and a genetic analysis should be made.

Points of discussion: NPHP is classified into three subtypes according to clinical manifestations. Althou- gh this case is classified as juvenile NPHP in terms of the age of ESRD onset, the diagnosis was difficult because the echo findings indicated infantile NPHP. Three cases with the same mutation have been reported, two of which are brother and sister who both developed ESRD at the age of 11 years, and the sister had visceral inversion. The third patient developed ESRD before the age of 2 years and

had comorbid liver fibrosis. These cases show that even with the same genetic mutation, different phenotypes may be expressed, which suggests the possibility of functional modification by other factors.

keywords: Infantile nephronophthisis (NPHP type 2), inversin (INVS) gene, renal enlargement, metabolic acidosis, tubular proteinuria

P2-4

A Follow-up family study from a woman with combined MYH9 and PAX6 mutations

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Backgrounds: Previously we reported the first case of a woman diagnosed with Fechtner syndrome and aniridia caused by combined MYH9 and PAX6 mutation. MYH9 mutation caused her renal insufficiency (first presented with proteinuria and now requires hemodialysis) and thrombocytopenia, while aniridia resulted from the PAX6 mutation. In this study, we report a follow-up study of her children.

Case: A son and a daughter of a combined MYH9 and PAX6 mutated mother came to the pediatric nephrology clinic for genetic evaluation as these genes above are inherited autosomal dominant. They were from different fathers. Her 11-year-old son had a history of unexplained thrombocytopenia immediately after birth requiring transfusions, and no further evaluation was done as his mother requested discharge. Her 6-year-old daughter was diagnosed with aniridia and nystagmus. The ophthalmologists who treated the daughter suspected PAX6 mutation. For hereditary nephropathy evaluation, blood and urine laboratory exams, hearing and eye evaluation were done in these children. The son had thrombocytopenia without clinical features of bleeding tendency, microscopic hematuria, and non-nephrotic range proteinuria were detected with preserved renal function. Glaucoma was suspected in the eye exam and planning for further evaluation. He had no abnormality in the hearing test. Angiotensin II receptor blocker was initiated for proteinuria control. The daughter had no abnormalities in blood, urine and hearing test. Sanger sequencing test of known maternal mutation of MYH9 and PAX6 was done in both children. The son had heterozygous MYH9 mutation from the mother and normal PAX6 gene. In comparison, the daughter had heterozygous PAX6 mutation from the mother and normal MYH9 gene. The daughter is planning to evaluate abdominal ultrasonography for extraocular manifestation of PAX6 mutation.

Conclusions: We present follow-up familial genetic evaluation from combined MYH9 and PAX6 mutated women. With the era of active genetic evaluation, offspring of known hereditary nephropathy patients should be concerned for early diagnosis of hereditary nephropathy for adequate and early inter-

vention to preserve kidney function.

Points of discussion: 1. When to consider the genetic test in children of hereditary nephropathy parents? 2. What capability should pediatric nephrologists possess in the era of genetic tests getting more available in practice?

Keywords: MYH9, PAX6, hereditary nephropathy

P2-5

Clinical and Pathological Investigation of Oligomeganephronia

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Backgrounds: Oligomeganephronia (OMN) is a rare subtype of congenital anomalies of the kidney and urinary tract, characterized by decreased number and compensatory hypertrophy of the nephron. Hyperfil- tration and excessive stress could cause glomerular injury and glomerulosclerosis, with renal failure typically occurring in adulthood.

Case: Ten patients pathologically diagnosed with OMN between 2013 and 2020 were retrospectively investigated. Eight of the ten cases were identified by urinary screening, and the remaining two cases were detected by incidental blood examination. For each of them, percutaneous ultrasound-guided renal biopsy was conducted once and diagnosed by one patholo- gist. The data were presented as the median ± interquartile range, and statistical significance was set at $p<0.05$. The age at diagnosis was 14.1 years, the male-to-female ratio was 6:4, and only four cases were born with low birth weight. The estimated glomerular filtration rate was 62.2 mL/min/ 1.73 m2 and the amount of proteinuria was 0.33 g/gCr. The pathological data showed glomeruli diameter of OMN patients was significantly larger (217 vs 155 μm, $p< 0.001$) than the control group, and the number of glomeruli of OMN patients was lesser (0.89 vs 2.75 / mm2, $p <0.001$) than the control group. Nine patients were treated with renin-angiotensin system (RAS) inhibitors, following which proteinuria successfully decreased or disappeared (from 0.33 to 0.080 g/gCr, $p<0.05$).

Conclusions: Renal dysfunction was observed in all patients at the time of renal biopsy. Although few symptoms presented in OMN, urine screening system was a major opportunity to reach the diagnosis of OMN.

Points of discussion: One of the problems regarding OMN is that the pathological diagnosis is relatively subjective and there is no obvious cutoff for the diagnosis of this disease. The percentage of OMN cases with genetic diseases is also unclear. Correction of glomerular hyperfiltration or hypertension might have reno-protective effects in OMN.

Keywords: Oligomeganephronia, congenital anomalies of the kidney and urinary tract, renal hypoplasia, screening

P2-6

A case of infantile nephrotic syndrome associated with retinal dystrophy

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Backgrounds: Infantile nephrotic syndrome (NS) is characterized by massive proteinuria and hypoalbuminemia presents between the age of three months and one year. More than 80% of congenital or infantile NS are caused by pathologic variants of NPHS1, NPHS2, NPH3, WT1, or LAMB2 genes. Pierson syndrome is an autosomal recessive disorder caused by a mutation in the LAMB2 encoding laminin $\beta 2$ peptides of laminin $\alpha 5\beta 2\gamma 1$ trimer, one of the major components of the glomerular basement membrane, and characterized by microcoria, congenital NS, and neurological problems.

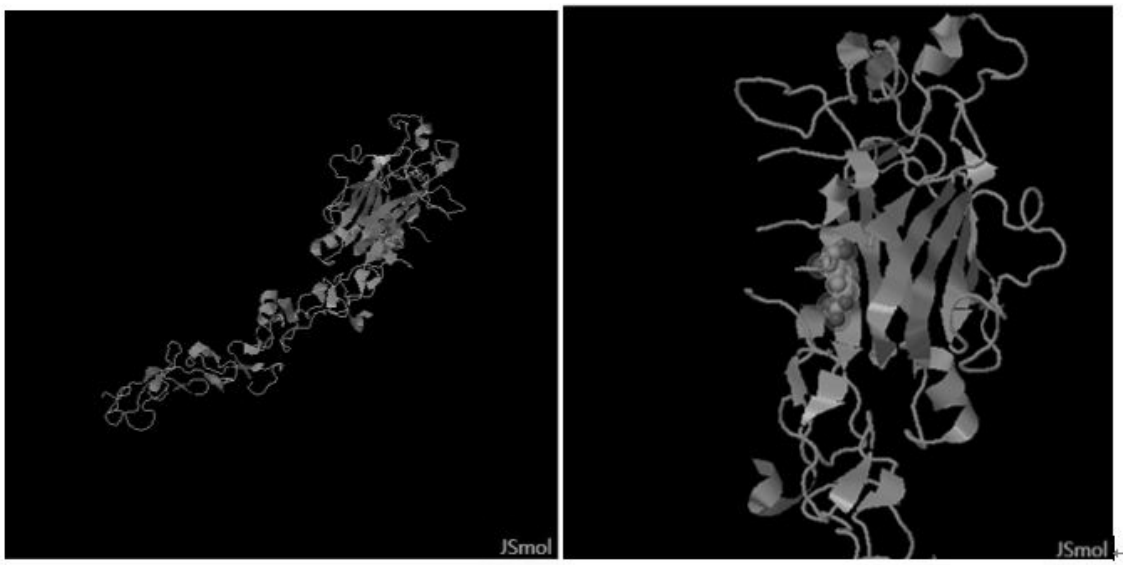
Case: A 6-month-old girl visited Seoul National University Children's Hospital with hyponatremia, proteinuria, and hypoalbuminemia along with urinary tract infection. She looked healthy without accompanied anomalies except for an odd eye. Her light reflex was prompt in both eyes without microcoria. Her height, body weight, blood pressure and laboratory findings were as follows; 69.7cm (75-90p), 8.3 kg (50-75p), 135/93 mmHg, serum BUN 41 mg/dL, serum /creatinine 0.27 mg/dL, serum albumin 1.4 g/dL, urine protein/creatine 17.01 mg/mg. While generalized edema was not prominent, albumin infusion resulted in weight loss of 1.7kg. There was no cystic lesion on the kidney ultrasound. A kidney biopsy was interpreted as mild diffuse proliferative glomerulonephritis with acute tubulointerstitial nephritis. To ameliorate the tubulointerstitial nephritis and unsure of genetic causes, she was treated with oral corticosteroid and captopril, followed by cyclosporine, but her proteinuria did not improve. Ophthalmologic examination showed bilateral hypopigmented fundus with incomplete vascularization. Genetic test revealed compound heterozygous variants in LAMB2 (from her father, and a variant of uncertain significance (VUS) from her mother). At 13 months of her age, she is well- being despite nephrotic range proteinuria. Her height, body weight, blood pressure and laboratory findings were as follows; 75.8cm (50-75p), 10.2 kg (75-90p), 131/ 70 mmHg, serum BUN 24 mg/dL, serum creatinine 0.41 mg/dL, serum albumin 3.4, urine protein/creatine 7.30 mg/mg.

Conclusions: Pathologic variants in LAMB2 have a broad spectrum of phenotypes from severe Pierson syndrome along with neurological deficit to isolated NS. If VUS of this patient is pathogenic, as predicted in silico, this can be another case of Pierson syndrome.

Points of discussion: How can we determine the pathogenicity of a VUS in a clinically compatible case? What is the best treatment for this case?

Keywords: Infantile nephrotic syndrome, LAMB2, Pierson syndrome

Figure. 3D visualization of LAMB2 protein with NM_002292.4:c.173G>A variant



PE-1

Rare Case of PR3-ANCA-Associated Vasculitis In a Child Manifested as C3 Glomerulonephritis

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Backgrounds: ANCA-associated vasculitis is a rare systemic vasculitides in children, especially PR3-ANCA associated vasculitis. Renal involvement of PR3-ANCA associated vasculitis manifested as C3 glomerulonephritis is rarely reported.

Case: A 13-year-old boy was presented to our hospital due to repeated gross hematuria for 3 years with recurrence for 3 days, accompanied with non-nephrotic proteinuria. The seizure frequency increased from 1-2 times per year to once a month and routine urine tests were normal during the interictal period. He had a previous history of allergic rhinitis and denied family history of kidney disease. External renal biopsy at 2 years after onset indicated focal sclerosing glomerulonephritis with renal tubular interstitial damage and no immune complex deposit, electroscope revealed glomerular segmental shrinkage, fusion of epithelial foot processes and neither of glomerular basement membrane thickening or electron dense deposit. There is no pathogenic gene variant detected by whole exome sequencing. Laboratory assessments at admission showed proteinuria (0.287g-0.535g/24h), glomerular hematuria (+++/HP, malformed erythrocytes 3.2□10^6/mL, G1>5%), elevated PR3(49U/L) and normal SCr (74 umol/L, eGFR 132ml/min/1.73m^2), ASO(98.10Ku/L, reference range 0~160), C3(0.88g/L, reference range 0.77~1.95), C4(0.26 g/L, reference range 0.07~0.40), MPO(□3U/ml), p-ANCA(-), c-ANCA(□). PPD-test, CMV DNA, EBV DNA and hepatitis virus test were negative. Renal ultrasound was also normal. Pathology consultation found occasional subepithelial deposition in Masson staining and nugget deposits in glomerular mesangial area and occasional subepithelial hump like dense deposits by electron microscope. Repeated renal biopsy show glomerular mild mesangial proliferation with predominant C3 deposits in glomeruli. Immunofluorescent test comfirmed that C4d was negative. Repeat testing found the level of C3 and C4 declined. With clinical diagnosis of C3 glomerulonephritis, the patient received ACEI treatment. At 7-month follow-up, microscopic hematuria still existed with C3 and C4 further declined (C3 0.69g/L, C4 0.15g/L). Based on persistence of 2 times increasing of PR3 antibody titer, diag-

nosis was corrected as PR3-ANCA associated vasculitis. Additional prednisone was initiated at 1 mg/kg/d, combined with mycophenolate mofetil 0.5g q12h. Two months after treatment, PR3 turned negative and prednisone was gradually reduced. At 13-month follow-up, the patient had no recurrence of gross hematuria, with negative urine protein and normal renal function.

Conclusions: Gross hematuria is common in pediatrics, vigilance of rare causes is necessary when the etiological diagnosis is not clear and therapeutic effect is not satisfactory. In this case, PR3-ANCA associated vasculitis is the potential cause of the child manifested as repeated gross hematuria and C3 glomerulonephritis. Both dynamic monitoring of complements and ANCAs and repeated renal biopsy are essential for explicit diagnosis. Low dose glucocorticoids with mycophenolate mofetil is effective.

Points of discussion: 1. What are the common kidney diseases and rare causes in children with renal histopathological manifestation of C3 deposition? 2. What is the difference in clinical and pathological features of renal involvement between PR3-ANCA and MPO-ANCA associated vasculitis in children?

Keywords: hematuria;ANCA;vasculitis;C3 glomerulonephritis

PE-2

A Case of Kikuchi-Fujimoto Disease associated with Lupus nephritis in a 10-Year-Old boy

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Backgrounds: Kikuchi-Fujimoto Disease (KFD) associated with systemic lupus erythematous in children are very rare, with only 1 case reported in China and only 15 cases abroad.

Case: A 10-year-old boy presented to hospital for a rapidly enlarging cervical lymph node, fever and rash, anti-infection treatments were ineffective. PE: Temperature 37.8-39.6℃ (irregular fever). Scattered erythematous maculopapular rashes were visible on his cheeks, pinna, and arms. Multiple bean-sized lymph nodes could be found on his neck, armpits and groin. The liver was slightly enlarged, 2 cm below the ribs was palpable, and the texture was soft. Laboratory tests: Urine protein (±), URBC (1+), 24h-Upro 0.109g, Blood WBC 3.70*109/L, Neut 2.2*109/L, Hb 92g/L, CRP 3.00ng/ml, ALT 101U/L, ESR 71 mm/h, anti- dsDNA 56.10IU/ml, ANA 65.00U/ml, C3 0.4g/L, Coombs test IgG(+). Ultrasound revealed the bilateral lymphadenopathy. Lymph node biopsy suggested histicytic necrotizing lymphadenitis and no hematoxylin bodies were found. Renal biopsy revealed Lupus nephritis III(A). We diagnosed with Kikuchi-Fujimoto Disease (KFD) with lupus nephritis. SLEDAI score was 8 points (2 points for low level of complement, 2 points for high level of anti-dsDNA, 4 points for hematuria). After treated with prednisone and mycophenolate mofetil, the fever, lymph nodes and rash of the child gradually subsided. After 4 months of treatment, the child's condition was stable and did not recur with SLEDAI score was 0 points.

Conclusions: Although KFD is a self-limited disease, it can be associated with or progresses to autoimmune diseases, of which SLE is the most common. There- fore, children diagnosed as KFD should pay attention to complete the SLE-related examination and follow up for at least 2 to 3 years. For children with HFD complicated with lupus nephritis, timely treatment with glucocorticoid and immunosuppressants will improve rapidly.

Points of discussion: 1. How to distinguish SLE from HFD early? 2. Treatment of KFD associated with lupus nephritis in children.

Keywords: Kikuchi-Fujimoto Disease, systemic lupus erythematous, children

PE-3

A case of acute kidney injury with systemic inflammation caused by TAFRO syndrome

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Backgrounds: TAFRO syndrome is a variant of idiopathic multicentric Castleman disease (iMCD) that has been recently recognized in Japan. It is a systemic inflammatory disorder characterized by a group of symptoms: thrombocytopenia (T), anasarca (A), fever (F), reticulin fibrosis in bone marrow (R), and organomegaly (O). TAFRO syndrome occurs mainly in middle-aged and elderly, and until now, only a few young patients have been described. We herein report a case of an adolescent who has been diagnosed as TAFRO syndrome, successfully treated with anti-IL-6 receptor antibody (Tocilizumab).

Case: A 14-year-old girl with a weight gain of 14 kg, generalized edema, and decreased urine output was referred to our center. She initially presented with fever and abdominal pain, and under the impression of pelvic inflammatory disease, she was treated with intravenous antibiotics. However, fever persisted and thrombocytopenia, ascites, and pleural effusion developed. As creatinine elevation and proteinuria worsened, she was treated with methylprednisolone and cyclosporine A, suspecting glomerulonephritis. Because kidney failure progressed, she was transferred to our hospital. Laboratory tests showed anemia, thrombocytopenia, hypoalbuminemia, elevated C-reactive protein (CRP), and increased BUN/Cr (86 mg/dL and 1.41 mg/dL). Autoantibodies including FANA, anti-ds DNA Ab, and antiphospholipid Abs were all negative. IL-6 level was moderately elevated (24.6 IU/mL). Imaging studies showed large ascites and multiple lymph node enlargements. For severe anasarca and kidney dysfunction, intermittent hemodialysis was started. Bone marrow biopsy revealed an increased number of megakaryocytes and mild myelofibrosis. Kidney biopsy showed features of thrombotic microangiopathy and lymph node biopsy was consistent with Castleman disease, hyaline-vascular type. Under the diagnosis of TAFRO syndrome, immunosuppressive therapy (steroid and cyclosporine A) was initiated, however, fever, thrombocytopenia, and CRP elevation persisted. For the second-line therapy, anti-IL-6 receptor antibody, Tocilizumab was administered every 2 weeks, and her clinical features and laboratory results

showed improvements. After a month, cyclosporine was discontinued, and corticosteroid was slowly tapered.

Conclusions: To our knowledge, this is the first report of adolescent histologically diagnosed with TAFRO syndrome in the Republic of Korea. The patient did not respond well to conventional immunosuppressants, but she was successfully treated with Tocilizumab. Though TAFRO syndrome is rarely reported in adolescents, further characterization of clinical and laboratory features is needed for a better understanding of the disease entity.

Keywords: Acute kidney injury, Castleman disease, Myelofibrosis, ascite

PE-4

Poststreptococcal glomerulonephritis mimicking membranoproliferative glomerulonephritis

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Backgrounds: Poststreptococcal glomerulonephritis (PSAGN) is a common glomerular disease in children and is generally not difficult to diagnose. However, a renal biopsy may be performed for diagnostic evaluation if an abnormal urinalysis or hypocomplementemia persists.

Case: A 7-year-old boy was presented to his primary care physician with gross hematuria. The patient had hematuria, proteinuria, low C3 level, and elevated antistreptolysin O level and was followed up for PSAGN. He had no hypertension, oliguria, or edema. However, hematuria and proteinuria persisted for 3 months, and the complement level did not normalize; therefore, he was referred to our hospital for evaluation by renal biopsy. Ninety-nine glomeruli were obtained, all of which were enlarged and had numerous neutrophil and monocyte infiltrates. There were no crescentic glomeruli. Masson's trichrome staining revealed a mass-like structure (hump), and periodic acid silver-methenamin staining revealed a double basement membrane. Fluorescence staining demonstrated granular deposits of C3, but it was negative for IgG. Electron microscopy revealed a rather thin glomerular basement membrane with scattered hump-like subepithelial deposits and intramembranous deposits. Based on histologic findings, we considered PSAGN; however, there were also findings suggestive of membranoproliferative glomerulonephritis (MPGN), such as doubling structures and intramembrane deposits, and we started steroid pulse therapy. After three courses of pulse therapy and posttherapy with PSL, the proteinuria disappeared 4 months after its onset. After the pulse therapy, the PSL was tapered. Persistent microhematuria also resolved after 1 year, and PSLs were discontinued simultaneously. The patient remained in remission for 5 years after the completion of treatment.

Conclusions: Renal biopsy, especially electron microscopy results, did not rule out MPGN or dense deposit disease (DDD), but nephritis symptoms resolved rapidly after the completion of steroid pulse therapy. The possibility that the histologic changes caused by MPGN/DDD were masked by those caused by PSAGN was also considered, but the disease course suggested the history of PSAGN.

Points of discussion: 1. What does pathology predict about how the disease progresses? 2. When should kidney biopsy be performed in the presence of persistent hypocomplementemia?

Keywords: Poststreptococcal glomerulonephritis; membranoproliferative glomerulonephritis; dense deposit disease; double basement membrane

PE-5

IgA nephropathy following COVID-19 vaccination requiring immunosuppressant therapy: A case report

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Backgrounds: Previous studies have reported many cases of new-onset glomerulonephritis after COVID-19 vaccination. Nevertheless, pediatric cases of glomerulonephritis after COVID-19 vaccination are limited. In addition, the use of combination therapy involving steroids and immunosuppressants to treat IgA nephropathy after COVID-19 vaccine is rare.

Case: A 12-year-old boy with a 2-day history of macrohematuria, fever, and right hypochondrium pain was referred to our hospital for further evaluation and treatment. Three days prior to presentation, the patient was vaccinated against COVID-19. The following day, his fever abated but macrohematuria persisted. He had no history of urinalysis abnormalities in the past. On admission, the patient had a normal body temperature with no significant findings on physical examination, except mild pain in the right hypochondrium. Urine and blood analyses showed significant proteinuria (a urine protein to creatinine ratio of 3.33 g/gCr), elevated urine red blood cells and casts, and mild renal dysfunction. There was no hypocomplementemia, and the autoimmune antibodies were negative. Kidney biopsy on day 14 revealed IgA nephropathy with diffuse mesangial proliferation, significant IgA deposits, and a 10% focal endocapillary proliferation. There was no crescent; however, a single glomerulus out of 38 glomeruli showed adhesions. One kur of methylprednisolone pulse therapy was administered, followed by combination therapy involving prednisolone, mizoribine, and angiotensin- converting-enzyme inhibitor. The patient's renal function promptly ameliorated, accompanied by a reduction in urinary protein, which returned to normal a month later.

Conclusions: Vaccination is an efficient strategy in managing the COVID-19 pandemic; this vaccination strategy will be continued in the future. Although the incidence of glomerulonephritis following COVID-19 vaccination is low, some reported cases have required treatment. Therefore, we should be attuned to the possibility of new-onset glomerulonephritis after COVID-19 vaccination. In addition, further case analyses should be performed to elucidate the mechanisms underlying the development of COVID-19 vaccine associated IgA nephropathy.

Keywords: IgA nephropathy, COVID-19 vaccine, immunosuppressant therapy

PE-6

Renal artery stenosis presenting as congenital nephrotic syndrome with hyponatremic hypertensive syndrome in a 2-months old Infant

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Backgrounds: Congenital nephrotic syndrome (CNS) is a rare condition often caused by genetic defects of glomerular filtration barrier. However, secondary causes such as congenital infection, maternal systemic lupus erythematosus, also may lead to this condition. On the other hand, hyponatremic hypertensive syndrome is a manifestation of severe hypertension related to renal ischemia, commonly from unilateral congenital renal artery stenosis in children. Over-stimulated renin-angiotensin-aldosterone system may cause proteinuria, polyuria and renal electrolyte loss. Here, we present a case of CNS along with hyponatremic hypertensive syndrome

Case: A 2months old boy was hospitalized with vomiting and general weakness. Physical findings showed high blood pressure (143/107mmHg). Laboratory tests revealed heavy proteinuria (Urine protein/creatinine ratio [uPCR] 107.25), hypoalbuminemia (2.6g/dL), hyponatremia (serum Na 123mmol/L), elevated renin/aldosterone (>80/206) and elevated serum creatinine (0.84 mg/dL). Workup for congenital nephrotic syndrome including congenital infection, search for associated anomalies, gene mutations was negative. Suspicious narrowing of Right renal artery with decreased vascular flow was shown in kidney Doppler sonography, and CT angiography revealed poorly visible renal artery proximal os. Right renal artery angiography was performed and right proximal and mid renal artery stenosis were found along with right proximal renal artery thrombosis. Balloon angioplasty was done without acute complication, which brought improvement of hypertension (BP 87/57mmHg), hyponatremia (serum Na 138mmol/L), proteinuria (uPCR 9.67), hypoalbuminemia (3.3g/dL), and high renin/aldosterone (10.64/22.3). His general condition was improved and ACEi, calcium channel blocker and beta blocker were used to control blood pressure. He was discharged with warfarin for thrombosis and anti-hypertensive medication of calcium channel blocker. In 3months, his proteinuria disappeared, and at last follow-up at his age of 23months, he was well-being with antihypertensive of beta blocker and normal blood chemistry.

Conclusions: Nephrotic syndrome may result from secondary to severe renovascular hypertension. In this case, accompanying hyponatremic hypertensive syndrome gave a clue to the underlying condition. Careful management of electrolyte imbalance, dehydration, and eventually correction of underlying structural problem can reverse the condition.

Points of discussion: What is the underlying cause of renal artery thrombosis in this baby? Is angioplasty a safe treatment for this young baby? How fast may we correct hyponatremia in this situation?

Keywords: Congenital nephrotic syndrome, Hypona- tremic hypertensive syndrome

PE-7

A novel hemizygous Trp245Cys GLA mutation in a 12-year-old Chinese boy with lupus nephritis and Fabry disease

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Backgrounds: Fabry disease(FD) is an X-linked lysosome storage disease caused by deficiency of the-alpha- galactosidase(α-Gal) enzyme. Systemic lupus erythematosus (SLE) is a chronic autoimmune disease with multisystem involvement and predominantly affects women of childbearing age.SLE and FD share similarities in the organs and may overlap. However, FD coexisting with lupus nephritis is extremely rare.

Case: A 12-year-old chinese boy was diagnosed with SLE because of fever, glomerular hematuria, a nephrotic range of proteinuria, hypocomplementaemia and positivity for antinuclear antibodies(ANA) and anti-double stranded deoxyribonucleic acid (anti- dsDNA). Light microscopy of kidney biopsy was characteristic of lupus nephritis (Class IV+V), whereas electron microscopy showed osmiophilic myelin-like bodies in the cytoplasm of glomerular podocytes. Immunofluorescence demonstrated deposition of IgG, IgA, IgM, C3 and C1q along the capillary loops and mesangial areas. The leukocytic α- GLA activity was abnormally low. Genetic analysis showed hemizygous for the c.G735C mutation in exon5 of GLA gene, which inherited from his mother and maternal grandmother who was heterozygous and asymptomatic. Hydroxychloroquine (HCQ) was discontinued based on renal pathology. Methylpre- dnisolone pulses and intravenous cyclophosphamide were commenced followed by maintenance therapy. The patient was also treated with angiotensin-converting enzyme inhibitors and angiotensin receptor blocker(ACEI/ARB). This treatment regimen brought about only partial im- provement. Enzyme replacement therapy (ERT, agalsidase alfa 0.2mg/kg intravenous administration every 2weeks) was initiated 2 months after diagnosis. Pro-teinuria levels were reduced to <500 mg/24h after 3 infusions.

Conclusions: To our knowledge, this case is the youngest patient with a novel mutation of SLE coexisting with FD had been reported. It is an important reminder of the role of renal biopsy as an indicator of Fabry disease coexisting with nephropathy. Genetic testing is especially necessary in male patients with lupus. Early intervention with ERT may help to relieve proteinuria.

Points of discussion: Several cases of concurrent Fabry disease and lupus nephritis have been reported, but the pathogenic association between these two diseases remains unclear and need to further investiga- tion. Despite ERT has demonstrated efficacy and safety on the treatment of FD, there are still some practical challenges such as the best regimen of glucocorticoid and immunosuppressants, the optimal time for repeat renal biopsy, the limited tissue penetration, the immunogenicity issues, the inconvenience and high cost of lifelong biweekly intravenous administra- tions.

Keywords: GLA mutation ; lupus nephritis ; Fabry disease

PE-8

A Noval Mutation of the TTC21B Gene Combined with TMA and Literature Review

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Backgrounds: TTC21B encodes the IFT139 protein, a vital component of the retrograde transport system within the primary cilia. The TTC21B mutation primarily leads to two kidney-related clinical phenotypes, NPHP and FSGS. TMA is a group of clinical syndromes caused by infection, complement mediation, malignant hypertension, etc. However, TMA has not been reported in the TTC21B gene mutation.

Case: At the age of five years and three months, the girl was hospitalized with oliguria and edema. Her primary clinical manifestation was malignant hypertension (189/132mmHg) and acute renal failure. Auxiliary examination showed oliguria, thrombocytopenia (59*10^9 /L), and serum lactate dehydrogenase increased by 1118(U/L). Renal biopsy pathology sho- wed widening of subcutaneous space, segmental basement membrane wormlike changes, arteriolar lumen stenosis, and C1q deposition, which diagnosed TMA. After treatment with nifedipine, nitroprusside, plasma exchange, and hemodialysis, the blood pressure of the child was controlled stably, and the hemolysis was alleviated. Whole-exome sequencing revealed that the TTC21B gene in the child had c.2461+1G>A(exon18) and c.1552T>C (exon13) compound heterozygous mutations. The former is a splicing mutation, and the latter is a missense mutation. The mother had a heterozygous mutation at c.2461+1G>A, and the father did not have genetic testing. The child had no family history of kidney disease, and her mother's first child was a twin. One fetus is arrested for developing in the seventh month of pregnancy, and the other fetus gave birth naturally, but there was no vital sign after birth. C.2461+1G>A is a new mutation, and c.1552T>C (p.Cys518Arg) has been reported. According to the American College of Medical Genetics and Genomics (ACMG) standards and guidelines, both variants could be classified as “Likely pathogenic” (PVS1+ PM2 and PS1+PM2+PP3, respectively). The literature review found more than 50 cases of TTC21B mutation in children. Most of them had short phalanx, combined damage liver and kidney, and developed into ESRD in childhood. Among them, NPHP is the most common phenotype, accounting for 56%, and FSGS

phenotype accounts for 28%; The primary type of mutation is p.Pro209Leu, which often appears in Euro- pean and American countries. However, there were 10 cases of p.Cys518Arg mutation, which only appeared in children with China. There are 8 cases of NPHP phenotype, 1 case of CHD, and 1 case of proliferative glomerulosclerosis in p.Cys518Arg mutation. Most have hypertension, proteinuria, and progressive renal failure in early childhood, similar to the child reported in this study. As for splicing mutations, 9 cases were reported, most of which had short phalanx or physical retardation, but the child reported in this study did not have such manifestations.

Conclusions: To sum up, we report a case of TMA with a novel TTC21B mutation (c.2461+1G>A), which expanded the gene spectrum and phenotype of TTC21B pathogenic mutation and explored the potential mechanism of renal injury. On the other hand, we summarized the pathological renal types of children with TTC21B mutation. We found the relationship between genotype and phenotype, that is, mutations in p.Cys518Arg are more likely to cause NPHP, and p.Pro209Leu mutation is closely related to FSGS.

Points of discussion: Regarding TMA in the child with TTC21B gene mutation, we believe that TTC21B gene mutation causes hypertension, thus inducing TMA, leading to kidney damage. At the same time, hypertension itself also affects the kidney.

Keywords: TTC21B gene, novel variant, TMA, children, hypertension, renal failure

TABLE 1 | Clinical phenotypes reported for TTC21B variants previously reported

Clinical phenotypes reported for *TTC21B* variants previously reported						
Number	Clinical phenotype	Gender	Age at onset(years)	Clinical symptoms	Age at ESRD	Mutation
1	NPHP	M	2	nephronophthisis, high myopia,nycatalopia	6	c.626C > T [Het] c.1525G > T [Het]
2	FSGS	M	3	primary ciliary dyskinesia, biliary dysgenesis,myopia, hypertension	4	c.626C > T [Het] c.1088-1G>C [Het]
3	proliferative glomerulosclerosis	F	16	elevated uric acid,hypertension	n	c.1552T > C [Het] c.2309A > G[Het]
4	NPHP	F	2	hypertension,hematuria,dextrocardia	3	c.1552T> C [Het] c.1231C> T [Het]
5	NPHP	M	2	hypertension,FSGS	2	c.1552T> C [Het] c.1231C> T [Het]
6	NPHP	M	1	hypertension,dextrocardia,FSGS	3	c.1552T> C [Hom]
7	NPHP	F	4 months	proteinuria	6 months	c.1675-1G> T [Hom]
8	NPHP	F	6 months	hypertension,FSGS	1	c.1552T> C [Het] c.1327C> G [Het]
9	NPHP	M	1	hypertension,development delays	2	c.1552T> C [Het] c.530delA [Het]
10	NPHP	F	3	severe early-onset hypertension,proteinuria, LV hypertrophy, liver function test abnormalities, growth retardation	3	c.626C> T [Hom]
11	NPHP	M	1	severe early-onset hypertension	5	c.2500C>T [Hom]
12	NPHP	M	6	renal hypertension,laterality defects	NA	c.1656_1659del [Het] c.1552 T > C [Het]
13	Neonatal Cholestasis	F	1	hyperlipidemia, chronic renal disease,neonatal cholestasis	NA	c.1656_1659del [Het] c.1552 T > C [Het]
14	Complex CHD	M	6	transposition of great arteries, ventricular septal defect, pulmonary stenosis,patent ductus arteriosus	NA	c.2322 + 3A > G [Het] c.349 T > C [Het]
15	Complex CHD	M	10	lateraldouble outlet of right ventricle, atrial septal defect, patent foramen ovale,pulmonary stenosis,laterality defects	NA	c.1464 + 2 T > C [Hom]
16	uncertain	F	2	laterality defects	NA	c.1464 + 2 T > C [Het] c.1625G > T [Het]
17	uncertain	M	4	laterality defects	NA	c.1799 T > A [Het] c.2071G > A [Het]
18	FSGS	F	4	myopia	6	c.626C> T [Het] c.1276C> G [Het]
19	FSGS	M	6	myopia	8	c.626C> T [Het] c.1276C> G [Het]
20	FSGS	F	12	hypertension	14	c.626C> T [Hom]
21	FSGS	M	8	myopia,hypertension	8	c.626C> T [Hom]
22	FSGS	F	17	myopia	NA	c.626C> T [Hom]
23	NPHP	M	7	physical retardation,hypertension	8	c.2211+3 A>G [Hom]
24	NPHP	F	10 months	hypertension	1	c.1552T>C [Het] c.1456dupA [Het]
25	NPHP	F	3	hypertension, hypertensive heart disease, heart failure, renal insufficiency, dextrocardia	4	c.1552T>C [Het] c.752T>G [Het]
26	FSGS	F	2	infant with brachydactyly, nephrotic-range proteinuria, renal tubular acidosis	2	c.626C>T [Het] c.40T>C [Het]
27	FSGS	M	early infantile period	renal cysts of various sizes, several small liver cysts, dilated bile ducts,polyuria with urinary concentration defects	1	c.1685A>G [Het] c.2569G>A [Het]
28	FSGS	F	15	hypertension	15	c.626C> T [Hom]
29	FSGS	F	15	hypertension	27	c.626C> T [Hom]
30	FSGS	F	18	hypertension	27	c.626C> T [Hom]
31	FSGS	F	18	hypertension,cerebral aneurysm,deafness	26	c.626C> T [Hom]
32	FSGS	M	9	hypertension,cerebral aneurysm	16	c.626C> T [Hom]
33	FSGS	M	18	hypertension,tubulointerstitial fibrosis	27	c.626C> T [Hom]
34	FSGS	F	16		17	c.626C> T [Hom]
35	NPHP	M	14	hypertension,severe scoliosis	20	c.626C> T [Hom]
36	NPHP	F	11	bilateral hiposteotomy	12	c.626C> T [Hom]
37	NPHP	F	10	hypertension	14	c.626C> T [Hom]
38	NPHP	M	11	hypertention, elevated liver enzymes	11	c.626C> T [Hom]
39	NPHP	M	8 months	hypertension,right moderate hydronephorsis	1.5	c.626C > T [Het] c.450G > A [Het]
40	NPHP	M	NA	hypertension	2.5	n
41	Bardet-Biedl syndrome	M	neonate	neonatal respiratory failure,renal dysplasia expands	NA	c.1846C > T
42	NPHP	M	9	polycysts,tubulo interstitial damage	8	c.1685A > G [Het] c.2569 G > A [Het]
43	NPHP	M	1	glomeruloscrelosis,tubulointerstitial damage	1	c.379 G > A [Het] c.2992_2994del [Het]
44	JATD	M	7	retrognathia, developmental delay	NA	c.2758-2A > G [Het] c.3857 T > C [Het]
45	JATD	NA	8	Narrow thorax, polydactyly left hand and foot,brachydactyly, shortened long bones, short stature	8	c.268_269 insTAGA [Hom]
46	NPHP	NA	infant	mental retardation, hepatic fibrosis	1.5	c.626C>T [Het] c.1654-7delTGTC [Het]
47	NPHP	NA	NA	primary sclerosing cholangitis	7	c.448T>C [Het]
48	NPHP	NA	NA	primary sclerosing cholangitis,vesicoureteral reflux	2	c.3264-3C>G [Het]
49	NPHP	NA	infant	short phalanges,hepatic fibrosis	2	c.2758-2A>G [Het] c.626C>T [Het]
50	NPHP	NA	NA	hypertention, chondrodysplasia,Bell's palsy	8	c.1231C>T [Het] c.3923A>G [Het]
51	NPHP	NA	NA	gastrointestinal tract malformation, situs inversus, polydactyly,polysplenia	3	c.264_267dupTAGA [Hom]
52	NPHP	NA	NA	liver fibrosis	2	c.626C>T [Het] c.1240G>T [Het]
53	NPHP	NA	NA	liver fibrosis, cone-shaped epiphysis	3	c.626C>T [Het] c.2868+IG>T [Het]
54	NPHP	NA	NA	situs inversus, hepatopathy	10	c.626C>T [Het] c.3923A>G [Het]

F, female; M, male; CHD,congenital heart disease; NA, not available.

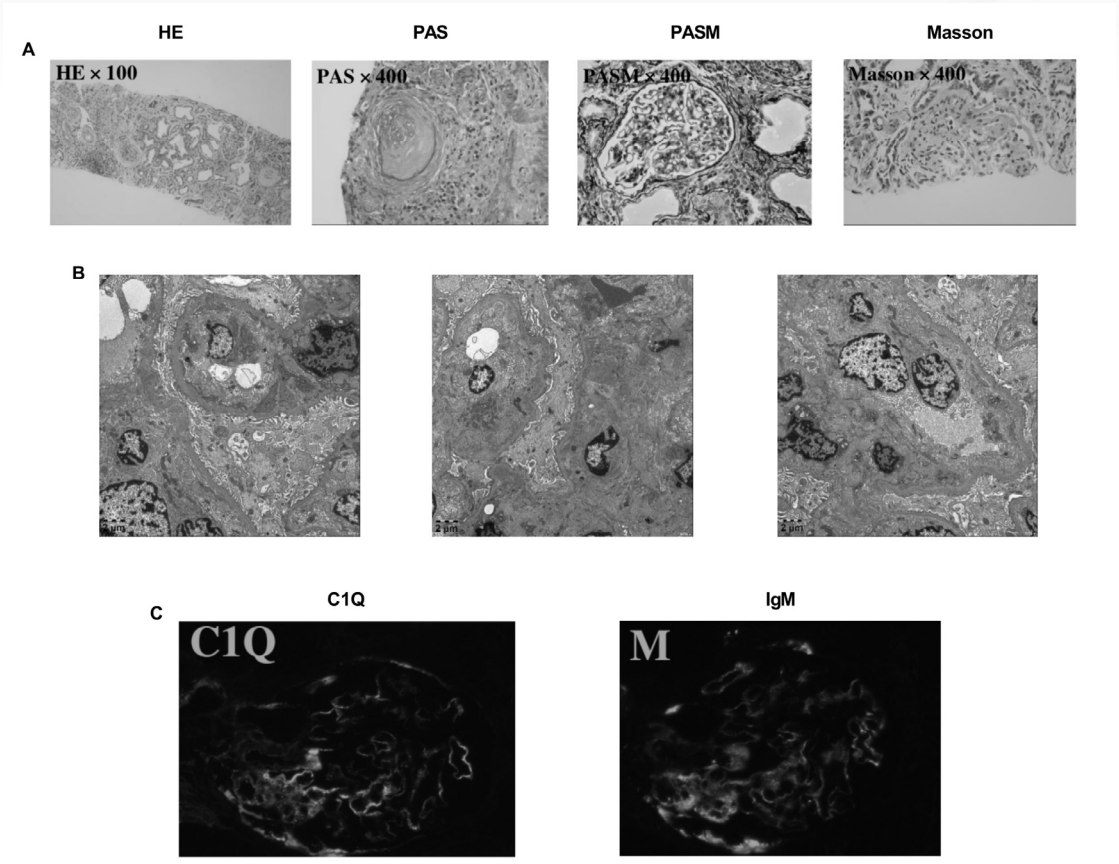

FIGURE 1 | (A) Kidney histological examination of the patient showed the pathological manifestations of hypertension. **(B)** Electron microscopic results of the patient supported TMA. **(C)** Results of immunofluorescence of the patient indicated C1q (++) deposited in a comma shape along the mesangial region and capillary loops.

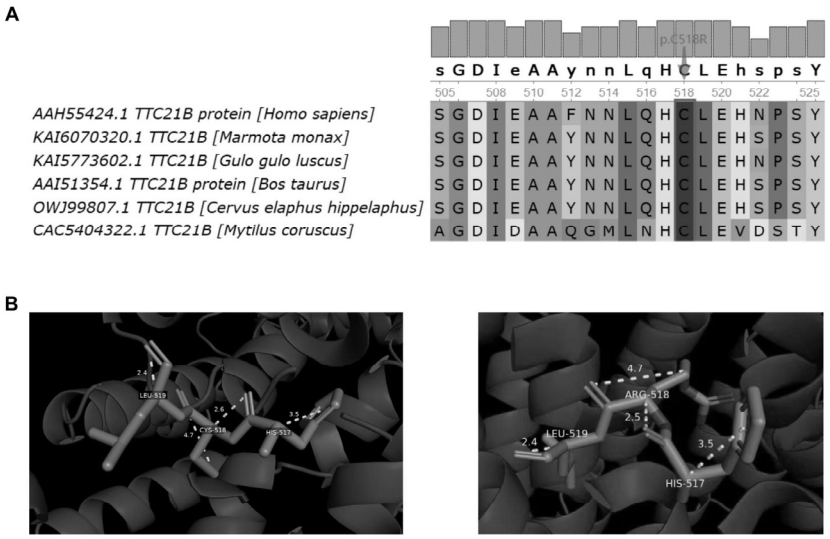

FIGURE 2 | (A) Conservative analysis of Pro518. **(B)** Predicted three-dimensional structure model of human *TTC21B* protein.

PE-9

Fanconi Syndrome as the First Manifestation of a Child with Kearn-Sanye Syndrome

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Backgrounds: Kearns-Sayre syndrome (KSS) is a rare mitochondrial cytopathy, KSS has a triad of features. The most common renal manifestation associated with Kearn-Sanye syndrome is proximal renal tubulopathy.

Case: A 3 years old girl had been diagnosed to be Fanconi syndrome at 3 years old when she felt lower extremities weakness in 2015. Routine blood chemistry showed hyperchloremia, hypokalemia, and metabolic acidosis. EMG displayed suspicious myogenic damage. She got better after symptomatic treatment in our hospital. The child developed growth retardation in 2017 (5 years old), whose height increased by about 1cm/year. The child had developed ptosis of the eyelids since 2020. No diurnal fluctuation was observed. The fatigue test was positive. The child was treated with steroids plus tacrolimus, but the treatment was ineffective. In July 2022(10 years old), edema, hyperglycemia, and acute heart failure developed in the patient. She was diagnosed for type 1 diabetes and III° atrioventricular block(AVB). Insulin was used for her diabetes and a pacemaker for her heart. The girl was admitted to our hospital again in October 2022. Her length was 112.5 cm (<3SD) and her weight was 17kg (<3SD). Clinical examination demonstrated severe eye ptosis and reduced eye fissures (about 4mm), limited eye movement in all directions. AG: pH 7.37, BE -7.3mmol/L; Lactate 1.8 mmol/L, sCr 61 μmol/L and cystatin C 1.28mg/L(eGFR 67ml/min.1.73m2), while her blood folic acid level was normal. Urinalysis revealed a generalized dysfunction of the proximal tubule with low-molecular-weight proteinuria and increased urinary Ca, and urinary P, urinary β2 MG (6.59mg/L), urinary α1 MG(36.6mg/L). Ultrasound of the urinary tract reveals small kidney stones. X-ray showed osteoporosis. Pure tone audiometry revealed high-frequency hearing loss. She had no retina problem or cognitive dysfunction. Cerebrospinal fluid test showed a increased protein level (2.23g/L) and a decreased of folic acid (27.2nmol/L(40-120)) . Besides, her head CT showed a widening of brain sulci and a narrowing of brain gyri. The muscle biopsy showed the presence of broken red fibers. Electron microscopy revealed an increased number of mi-

tochondria and suspicious disc-shaped mitochondria, and some mitochondria showed the formation of lattice-like inclusion bodies. The mitochondrial DNA sequencing showed a 7521-bp deletion mutation from the nucleotide position 7974 to 15,494. Cocktail treatment were given for treatment, and the symptoms of ptosis were relieved.

Conclusions: Fanconi syndrome is one of the onset manifestations of mitochondrial cytopathies. mitochondrial diseases such as KSS should be considered,if a child diagnosed as Fanconi syndrome who present with extrarenal manifestations such as ptosis. KSS can cause serious complications such as III°AVB and require active diagnosis and treatment.

Points of discussion: 1. How to improve the long- term prognosis of Kearn-Sanye syndrome? 2. Is there any better treatment for this patient?

keywords: Kearns-Sayre syndrome, Fanconi Syndrome

PE-10

Pediatric SRNS associated with LAMA5 mutation

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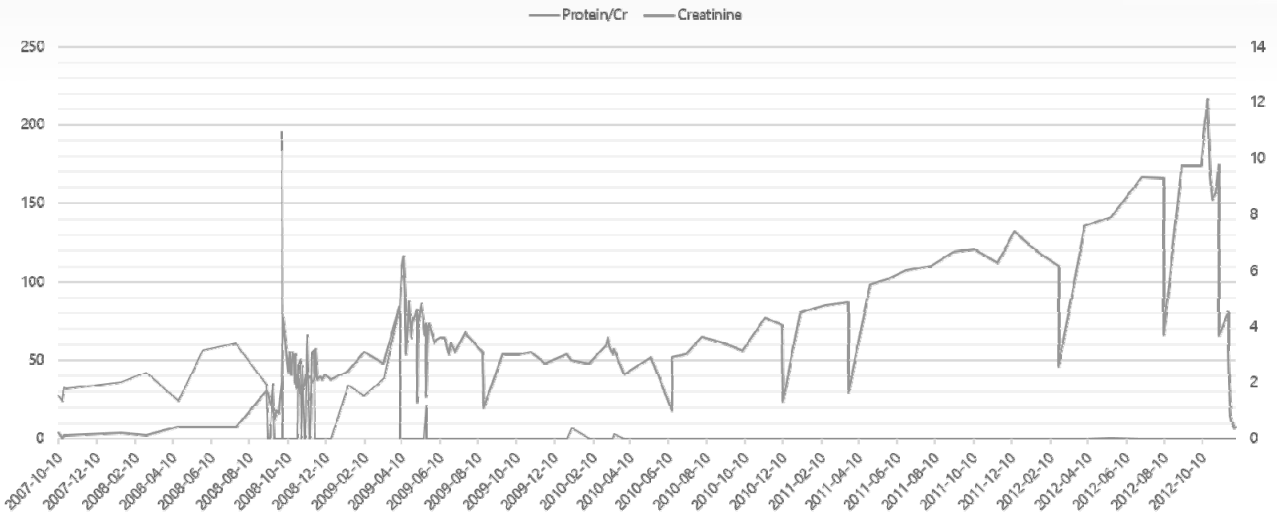
Backgrounds: Steroid-resistant Nephrotic Syndrome (SRNS) is a subtype of nephrotic syndrome characterized by proteinuria, hypoalbuminemia, and edema that does not respond to steroid therapy. Single gene pathogenic mutations have been implicated in up to 30% of pediatric SRNS, and over 70 genes have been reported to date. Of those recently discovered is LAMA5, which encodes the laminin-alpha-5 chain. Forming the laminin $\alpha 5\beta 2\gamma 1$ hetero-trimer, it is not only an essential component of the glomerular basement membrane (GBM) but also important for embryogenesis and immune modulation. Homozygous or compound heterozygous variants of LAMA5 have been identified to date in ten pediatric NS patients with variable phenotypes. These patients had onset of NS ranging from 3 months to 8 years. Response to therapy and renal outcomes varied from steroid sensitive NS to early end-stage kidney disease (ESKD). Biallelic truncating mutations of this gene were proven to cause SRNS recently.

Case: Here we present a case of infantile SRNS related to compound heterozygous variations of LAMA5 (c.3434G>A, p.Cys1145Tyr and c.6883C>T, p.Gln2295*). A 10-month-old female presented with eyelid edema and massive proteinuria without any extra-renal symptoms or family history. She was diagnosed with SRNS and renal biopsy revealed focal segmental glomerulosclerosis with widely effaced epithelial foot processes and "moth- eaten" appearance of GBM. She progressed to ESKD requiring dialysis at 3 years and 5 months of age, and received deceased-donor kidney transplant at 6 years of age. 4 months after transplantation, she developed EBV-related post-transplant lymphoproliferative disease (PTLD), which was treated with chemotherapy.

Conclusions: This case provides additive evidence that LAMA5 variants are related to SRNS.

Points of discussion: While our case has one missense and one truncating allele, her phenotype is similar to those with biallelic truncating variants, possibly because her missense variant alters splicing. Whether LAMA5 defects has played a role in vulnerability to PTLD is yet to be investigated.

Keywords: LAMA5, nephrotic syndrome, SRNS,


Protein/Cr
Creatinine
250
200
150
100
50
0
14
12
10
8
6
4
2
0
2007-10-10
2007-12-10
2008-02-10
2008-04-10
2008-06-10
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PE-11

THSD7A-Related Membranous Nephropathy as Paraneoplastic Glomerular Disease Secondary to Neuroblastoma

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Backgrounds: Malignancy is a rare cause of nephrotic syndrome in children and can be fatal if missed. Unlike in adulthood-onset tumor or pediatric hematologic malignancy, paraneoplastic glomerular disease secondary to solid tumor had been rarely reported in children.

Case: A 5-year-old boy was admitted with arthralgia for 3 months, edema of lower extremities and oliguria for 3 days. Urine protein of 3+, blood Hb of 82 g/L and ALB of 19 g/L were detected in outpatient department. No special family history. PE: P 125/min, R 34/min, pale skin, edema. Examination revealed: Urine RBC 0/uL, urine WBC 0/uL, urine protein 1.441g/24h; Blood WBC 5.2x10^9/L, Hb 82g/L, PLT 432x10^9/L; ALB 19 g/L, BUN 2.9 mmol/L, SCr 19 μmol/L, LDH 598 U/L; CHOL 9.8 mmol /L; D-dimer 11.38 mg/L, FIB 12.86 g/L; Ferritin 413 ug/L; Normal serum potassium, sodium, calcium, uric acid, ALT, AST, PT, APTT, RF, C3, C4, Coombs test, ANA, ANCA, ASO, PPD test, CMV/EB-DNA, HBsAg, etc. Ultrasound revealed a hypoechoic lesion in the right adrenal area. CT revealed the size of the mass was about 6.4×4.0×6.0cm^3, accompanied by multiple tubercles or masses in abdominal cavity and retroperitoneum. Further examination revealed blood NSE of 261.40 ng/mL (normal: <16.30), urine VMA of 228.09 μmol/L (normal: 24.9-70.2). Percutaneous puncture biopsy of adrenal mass revealed neuroblastoma. 3 months later, during autologous kidney transplantation, renal biopsy revealed membranous nephropathy stage I~II. THSD7A protein was detected in glomeruli by mass spectrometry. However, serum THSD7A antibody was (-). We took frozen serum (at the first visit) for reexamination of THSD7A antibodies, and the result was (+). In terms of treatment, symptomatic support such as albumin supplementation, diuresis and anticoagulation were given at the beginning, followed by high-risk-neuroblastoma chemotherapy (including high-dose CTX) after tumor diagnosis. The patient was also treated with glucocorticoid for NS (by other hospital), but without complete remission after 2 months. "Neuroblastoma resection + right kidney autotransplantation" was performed when operation conditions for neuroblastoma were met. Urine protein turned negative at the 3rd month. It has been 8

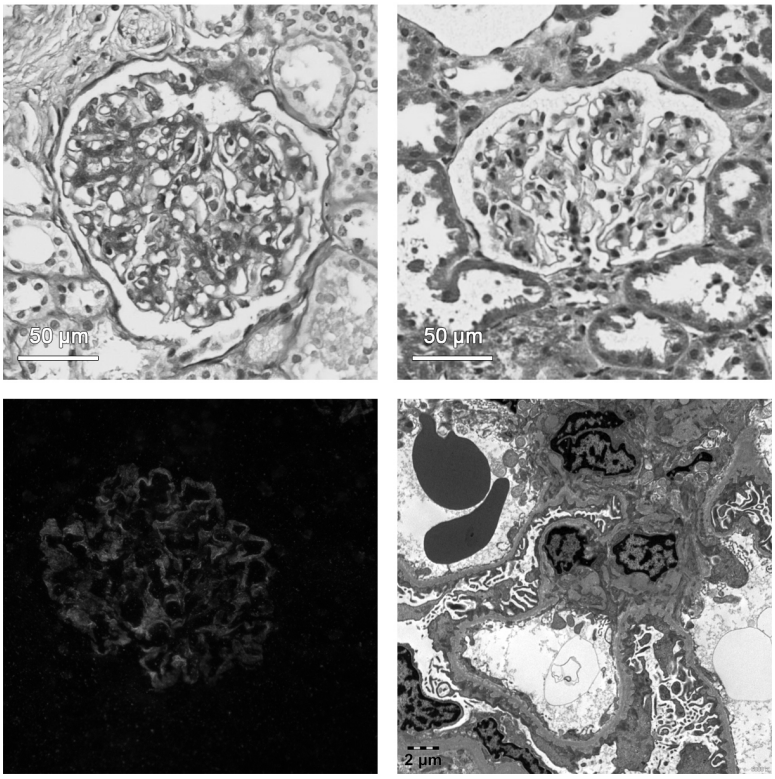
months after remission, while no recurrence of NS was observed. Tumor treatment is still continuing.

Conclusions: We report a case of NS secondary to neuroblastoma, which turned out to be anti-THSD7A- associated membranous nephropathy. Treatment is still focused on the primary tumor.

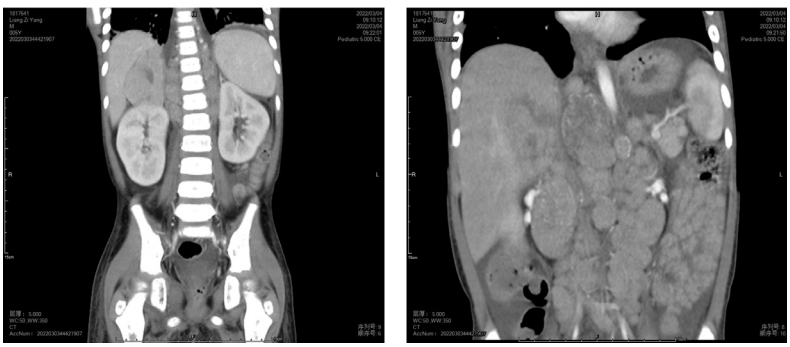
Points of discussion: 1. Screening strategies for tumor in nephrotic syndrome of children (but not adults); 2. Could immunosuppressive drugs in tumor chemotherapy be beneficial to NS remission?

Keywords: Paraneoplastic Glomerular Disease; Membranous Nephropathy; THSD7A; Nephrotic syndrome; Neuroblastoma

Renal Pathology (PAS, Masson, IgG, EM)



Abdominal CT



PE-12

A Rare Case of Proliferative Glomerulonephritis with Monoclonal IgG1-κ Deposition in a Child

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Background: Proliferative glomerulonephritis with monoclonal immunoglobulin deposits (PGNMID) is an extremely rare entity, especialy in children. The etiology are uncertain and lacks marked clinical manifestations. The pathological features are predominantly membranoproliferative glomerulonephritis, electron microscopy(EM) revealed nonorganized glomerular deposits and stained single heavy and light chain under immunofluorescence(IF), IgG3-κ subtype is most common.But this case is a rarer type of IgG1-κ PGNMID.

Case: The patient was a 14 year old girl with proteinuria fluctuated between 0.14-0.58g for 2 years,her serum albumin 45.89g/L, urine protein: 0.54g, Renal biopsy: IF: bright granular glomerular staining for IgG, IgG1,C3,κ-light chain diffusely distributed in the mesangial area , with negative IgM, IgA, C1q, and λ. Paraffin fluorescence: κ ++~+++;λ (-) LM: Mild proliferation of mesangial cells and mesangial matrix ; EM: granular electron dense deposits were present along mesangial and occasional hump-like deposits in the subepithelial. Meanwhile the rest of investigations like hepatitis B and C, ANA, C3、C4 and rheumatoid factor, bone aspiration biopsy ,serum and urine protein immunofixation electrophoresis and serum free light chain(SFC) assay were all within normal limits. ARB and seriod therapy continued after discharge, urine protein was negative at nearly 1 year follow-up.

Conclusions: A 14-year-old girl with proteinuria,mild mesangial proliferative nephritis as showed , positive mesangial area distribution of IgG, IgG1,C3,κ-light chain of IF,EM showed electron dense deposits with no-organized structure. Therefore, PGNMID IgG1-Kappa was clearly diagnosed.

Points of Discussion: PGNMID is characterized by the direct deposition of Mlg or its structural components in the kidney. It can be observed in a wide range of age, most frequently in patients in their 50s, but rarely in children. The gene sequencing has found heterozygous mutations in complement bypass related factors associated with PGNMID, which may lead to abnormal activation of the pathway. 1) Diagnosis & differentiation: renal biopsy has a key diagnostic value, which IF staining and EM finding

can help us distinguish other pathological types of MGRS, e.g., amyl-oidosis, light chain deposition disease, etc. Even if for children ,we should pay more attention to IF staining for κ、λ light chains, and if necessary, paraffin fluorescence to verify the reliability of κ、λ light chains staining. Furthermore, studies showed that there was an association between PGNMID and hematological malignancy, so we recommend a bone marrow aspiration biopsy in patients suspected of PGNMID, serum and urine protein electrophoresis, immunofixation, analyses of SFC should also be performed, which helps to establish and identify diagnosis. 2) about IgG subtype: In different subtypes of PGNMID, lgG3 is the most common for single lgG deposits, followed by lgG1, $\kappa > \lambda$. According report, the intensity of glomerular C3 deposition and the frequency of hypocomplementemia of C3 were higher in IgG3-PGNMID, IgG1 -PGNMID may have a higher correlation with extrarenal disease. These findings suggest that two subtypes differ in the mechanism of complement activation, finally lead to different pathology type. In this case, there were no evidence of hematological positivity or genetic abnormalities, ARB and hormone therapy worded well for her, and follow-up indicators turned to negative, so the lesion was considered mild as for the phenomenon, whether the special subtype IgG1-κ PGNMID associated with a better prognosis, it was still enigmatic and further follow-up and studies are needed.

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