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On behalf of ISPN bulletin editorial board I extend greetings to all of you! World Kidney Day 2024 was celebrated with great enthusiasm in our country. We bring the related news in this issue. ISPN members are training pediatricians and young nephrologists in various regions. Do read the related news in this issue. We also have an interesting case in the quiz section.

Do join us in Delhi for ISPNCON 2024 from 6th - 8th December at hotel Lalit.

I thank all the members of the *editorial board* who have worked hard to bring the issue, and also to all the members of ISPN for writing articles for this issue, all what matters the most is that we work as a TEAM.



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In this section we provide you important academic and administrative information from ISPN secretariate. In this issue we have compiled the activities conducted on World Kidney Day around India and important announcements.

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In this section we bring to you the activities conducted across various centres in India and announcement of upcoming events.



ADVANCING EQUITABLE
ACCESS TO CARE
AND OPTIMAL
MEDICATION PRACTICE

**KIDNEY
HEALTH
FOR ALL**



Kidney Health for all: Advancing equitable access to care and optimal medication practice

Dr Soumya Reddy

St John's Medical College Hospital, Bangalore

Chronic kidney disease (CKD) in low and low-middle-income countries (LMIC) have drastically worse outcomes compared to high-income countries (HIC). Although the causes of CKD are similar in both settings, children and adults in LMIC typically present late in the course of the disease with advanced CKD or kidney failure, have severe co morbidities, and have limited access to CKD care, especially kidney replacement therapy (KRT). The theme for World Kidney Day 2024, 'Kidney Health for all: Advancing equitable access to care and optimal medication practice', aimed to highlight the stark differences in the accessibility and delivery of kidney healthcare services between the two populations, and advocate for a comprehensive strategy to close the health gap.

Among vulnerable populations like children, the disparities are more glaring. It is reported that <10% of children in LLMIC who require dialysis receive it and even fewer have access to kidney transplants. A majority of children with kidney failure in these settings discontinue treatment or die due to inadequate pediatric nephrology centers, significant financial burden, limited health literacy, lack of a well-developed deceased donor program, and high caregiver burnout. The general lack of awareness regarding pediatric kidney disease among the community and healthcare personnel working at the grassroots level contributes to delayed referrals and increased risk of complications. Compared to HIC, the disparity in access to maintenance dialysis services is further compounded by the lack of universal health care coverage in most LLMICs forcing families to experience extreme financial and mental hardship. Limited availability of even essential medicines and exorbitant cost of disease-limiting therapies such as monoclonal antibodies and enzyme replacements leave patients and their families feeling helpless. Optimal medication practice will be a reality only when drugs are available, accessible, and affordable to everyone who needs them.

Addressing these challenges require a multifaceted tailored approach, including policy changes, healthcare infrastructure improvement, collaboration with HIC centers to create a support network, increasing community awareness and engagement, and establishing family support groups and education initiatives. By implementing these strategies, we can create an equitable kidney care system and a supportive environment that fosters holistic well-being and improves the quality of life and long-term outcomes of children with kidney disease.

IPNA World Kidney Day Contest 2024



The George Institute for Global Health India won the IPNA World Kidney Day Contest 2024 for their proposal on creating awareness about kidney health and screening for kidney disease among school children from a CKD endemic region in Andhra Pradesh.



Dr YSR KIDNEY RESEARCH CENTRE AND SUPER SPECIALITY HOSPITAL , PALASA

**WE CORDIALLY INVITE YOU FOR THE EVENT
WORLD KIDNEY DAY**



Kidney Health For All

ADVANCING EQUITABLE ACCESS TO CARE
AND OPTIMAL MEDICATION PRACTICE

SCREENING CAMP AND INFORMATIVE SESSION FOR
SCHOOL CHILDREN AND HEALTH CARE PROVIDERS



Dr BALAJI GUMMIDI
Senior Project Manager
The George Institute for Global Health



The George Institute
for Global Health India



KITE: Kidney Insight Through Education Screening Camps and Informative Sessions for School Children, and Health Workers

Event Title: KITE: Kidney Insight Through Education - Screening Camps and Informative Sessions for School Children, and Health Care Providers

Date: March 14th to 15th, 2024

Locations:

1. Kidney Research Innovation and Patient Assistance Centre (KRIPA) at Dr YSR Kidney Super specialist hospital and research centre, Palasa.

2. Zilla Parishad Govt. High School (Block level), Srikakulam District, Andhra Pradesh, India.

Overview: The World Kidney Day event, themed "KITE: Kidney Insight Through Education," aimed to raise awareness about kidney health and provide essential screenings and informative sessions for school children and health care providers. The event encompassed screening camps, information sessions, multiple activities for school children, launching of a Kidney Patient Assistance Centre, and workshops for community health workers. All these activities were conducted in Uddanam, a well-known chronic kidney disease hotspot.

Day 1: March 14th, 2024 - Dr YSR Kidney Super specialist hospital and research centre, Palasa

Event highlights:

Unveiling of Logo: The launch of the official World Kidney Day (WKD) 2024 theme and logo marked the beginning of local efforts to raise awareness about kidney health. The WKD 2024 focuses on "Kidney Health for All" was explained to promote equitable access to kidney care. The unveiled logo incorporates imagery related to kidneys and general health, signifying WKD's commitment to supporting those with kidney disease.

"Great news for kidney care in Uddanam region! We're providing free comprehensive kidney care services. Thanks to IPNA and the George Institute for this awareness session, people now know about these services. Don't forget, regular kidney check-ups are important for everyone". Dr Kuna Mohan Babu, Resident Medical Officer, Dr YSR Kidney Research Centre Super Speciality Hospital

Launch of patient assistance centre: A key achievement of the World Kidney Day program was the launch of the Kidney Patient Assistance Centre at the Government Kidney Super Specialty Hospital in Palasa, a known CKD hotspot in Uddanam region. This centre provides a lifeline for those battling kidney disease by offering emotional counselling, educational programs, resource materials, appointment scheduling assistance, financial aid support, and facilitated support groups. This comprehensive approach empowers patients to manage their condition, navigate the healthcare system, and connect with a supportive community, ultimately improving their quality of life.



“We're here to help people in Uddanam with kidney issues by offering all sorts of medical help like tests, medicines, and dialysis. Soon, we'll start kidney transplants, with all the right gear. If people take care and stay safe, their lives can get a lot better.” Dr Rakesh, Nephrologist, Dr YSR Kidney Research Centre Super Speciality Hospital



Launch of the Kidney Patient Assistance Centre at the Government Kidney Super Specialty Hospital in Palasa by hospital super intendent, Nephrologists and Senior Project Manager.

Information session for Health care providers: An educational training program targeting health care providers and allied medical staff from all levels of care in the Uddanam region was held. Led by experts in pediatrics, nephrology, nutrition, and public health research, the session aimed to improve their understanding of kidney diseases. With 84 participants, the program focused on equipping healthcare workers to identify early signs and symptoms of kidney disease, the importance of hydration, and emphasizing the importance of timely intervention. The program included nutrition management training and offered digital community outreach tools. Medical officers engaged in discussions about the management of people with kidney disease and provided updates on the latest guidelines and the importance of childhood screenings and management of urinary tract abnormalities.



Dr YSR Kidney Research Centre and Super Speciality Hospital Staff (Nephrologist, urologist and nutrition specialist interaction with medical officer and paramedical staff.

“On this World Kidney Day, we had an important discussion about taking care of our kidneys. We learned that it is important for everyone to get screened at regular intervals and to make sure that there are no issues with their urinary system. This can greatly affect their health as they grow up”.

Medical officer, Primary Health Centre



KITE: Kidney Insight Through Education Screening Camps and Informative Sessions for School Children, and Health Workers

Day 2: March 15th, 2024 – Zilla Parshad (Block level) Government High School, Rangoi village, Palasa. Day 2 of the World Kidney Day event kicked off with a vibrant screening camp at Zilla Parshad Government High School, Rangoi, Palasa, specifically designed for school children of 13 to 16 years group. A health team comprised of health volunteers, community health workers from The George Institute of Global Health, along with general practitioners and paediatricians conducted health screenings on over 55 participants. These screenings included urine analysis. To ensure accurate results, children were provided with sterile urine collection containers one day prior and instructed on how to collect amidst ream urine sample in a sterile manner properly. The collected samples were then analyzed for urinary parameters such as protein, glucose, ketones, pH, specific gravity, red blood cells (erythrocytes), and white blood cells (leukocytes). Height and weight measurements were taken, BMI was calculated, and questions were asked about hydration practices and family history of CKD to assess the overall health of the young attendees.



Urine analysis for the students participated in the screening camp.



Student screening camp measuring the height and weight.

IPNA World Kidney Day Contest 2024



Further engaging the children, the program included speech and drawing competitions on the themes of "Kidney Health for All" and "Our World, Our Health" These competitions garnered

Enthusiastic participation, with 55 registrations for the essay writing competition and 44 entries for the drawing competition.

To promote kidney health awareness from a young age, the event also incorporated educational materials emphasizing the importance of proper hydration and maintaining good kidney health. The educational aspect continued within formative talks delivered by nephrologists and public health specialists. These talks delved into critical topics such as kidney function, the importance of proper nutrition and hydration, the benefits of regular screenings, and effective management strategies for chronic kidney diseases. This multi-pronged approach ensured a fun and engaging environment where children not only received health screenings, but also gained valuable knowledge about kidney health.

"As teachers, it's important for us to know when someone is dehydrated or having a kidney problem. Understanding the signs and symptoms helps us teach our students and families about taking care of their kidneys" - Teacher at Zilla Parshad Government High School



Pediatric Nephrology Conclave and Dialysis Workshop at AIIMS, Bhopal



Conclusion:

The World Kidney Day event, "KITE: Kidney Insight Through Education," successfully fulfilled its objectives of raising awareness, conducting screenings, and providing informative sessions for both school children and healthcare providers. The launch of the Kidney Patient Assistance Centre marked a significant milestone in providing comprehensive support to kidney disease patients. The screening camp for school students and informative sessions for healthcare providers and children reinforced the importance of the role in spreading awareness and ensuring timely interventions, thus contributing to the overall improvement of kidney health in this region, well known as a CKD hotspot.

Acute Peritoneal Dialysis Hands-on Workshop at IGIMS, Patna

Dr. Anand Gupta, IGIMS, Patna

The first Pediatric Peritoneal Dialysis Hands-on workshop in the state of Bihar was organized by Dept of Pediatrics, IGIMS at Patna in association with ISPN on 16.06.24. Organizing Secretary was Dr. Anand Kumar Gupta, Organizing Co-Secretary were Dr. Amit Kumar and HOD Pediatrics Dr. (Prof.) Jayant Prakash. Lead instructor Dr. Sumantra Raut and other trainers Dr. Ranjeet Thergaonkar, Dr. Mritunjay Kumar, Dr. Arun Kumar and Dr. Anand Kumar Gupta with their cumulative efforts trained 40 residents of Pediatrics and Nephrology from different medical colleges of Bihar. Workshop included Hands on experience of stiff PD catheter insertion on Dummy-Tummy for each delegate with Baxter India being the collaborator.





Acute Peritoneal Dialysis Hands-on Workshop at RIMS, Ranchi

Dr Kiran Das

RIMS, Ranchi

The First Pediatric Peritoneal Dialysis Hands-on workshop in Jharkhand was organised by the Department of Pediatrics, RIMS at Ranchi in association with ISPN on 29.7.24. Organising Secretary Dr Kiran Das, HOD Pediatrics Prof Rajiv Mishra, Lead instructor Dr Sumantra Raut and other trainers Dr Pragya Pant, Dr Anand Gupta, Dr Rajan Kumar and Dr Pritha Naaz with their cumulative efforts trained 40 residents from Pediatrics and nursing staff from neonatology. The workshop included a hands-on experience of stiff PD catheter insertion on Dummy-Tummy for each delegate with Baxter India being the collaborator. The real-time result of the workshop was evident within a week when junior residents successfully did the 1st acute P DonanAKI child with complicated falciparum malaria.



Resident acute successfully performed 1st PD in a patient with falciparum malaria within a week of the PD hands-on workshop



Phase-1 CAPD rollout program throughout the state of West Bengal under PMNDT

Dr Sumantra Kumar Raut NBMC Darjeeling

The Department of Health and Family Welfare, Govt. of West Bengal has adopted the strategy to start a Continuous Ambulatory Peritoneal Dialysis (CAPD) program throughout the state free of cost to the patients (both adults and pediatrics) under PMNDP (Pradhan Mantri National Dialysis Program). Training programs for all tiers of HCWs (physicians, surgeons, critical care team, nurses, dialysis technologists, social workers, and also administrative staff such as deputy superintendent, assistant superintendent medical and nursing) were done phase-wise from October 2023 to January 2024 with the help of a team of Nephrologists, Pediatric Nephrologists, and Baxter team. CAPD connectology and catheter care were taught with the help of a dummy tummy. Five medical colleges in various zones of Bengal (NBMC Darjeeling, IPGMER-SSKM Hospital Kolkata, CMC Kolkata, NRSMC Kolkata, BMC Bardhaman) were selected for such training as per "hub and spoke" model. These centers will be selected for initiating new CAPD patients and managing complications. The different peripheral spoke centers were trained to maintain CAPD patients, their logistics, and planning for regular delivery of CAPD consumables to the patients. A 5-year-old girl already on CAPD was the 1st patient from the North Bengal area to avail of this facility. Once the CAPD program catches pace it will not only help the common people of Bengal and nearby with kidney failure, particularly the younger child in whom maintenance hemodialysis is logistically difficult in the periphery but will also reduce the load in the existing hemodialysis centers all over Bengal.



1st training session at NBMC, Darjeeling for phase-1 CAPD roll out program



3rd training session at CMC, Kolkata for phase-1 CAPD roll out program



IPNA- sponsored CME in JIPMER, Pondicherry

Dr Bobby Deepthi

JIPMER, Puducherry

An International Pediatric Nephrology Association (IPNA)-supported CME entitled 'Pediatric Nephrology in 2024: A primer and current best practice' was organized on February 10, 2024 at JIPMER, Puducherry. The organizing faculty included Dr Sriram Krishnamurthy, Dr Sudarsan K and Dr Bobby Deepthi. More than 150 delegates participated in the conference. 18 eminent pediatric nephrologists from institutes from all over the country participated as resource persons. A wide range of topics in pediatric nephrology such as frequently-relapsing/steroid-dependent nephrotic syndrome, steroid-resistant nephrotic syndrome, crescentic-glomerulonephritis, hematuria, hemolytic-uremic-syndrome, metabolic alkalosis, acute kidney injury, urinary tract infections, refractory-rickets, and hypertension were covered; and the educational activity was appreciated well by the delegates.



Faculty for the CME



Delegates and Faculty of the CME



Organizing team: Pediatric Nephrology in 2024: A primer and current best practice CME at JIPMER, Puducherry

Peritoneal dialysis teaching program

Dr Neha Agarwal, SMS Medical College, Jaipur

The Department of Pediatric Nephrology SMS Medical College, Jaipur conducted the first Peritoneal dialysis teaching program endorsed by the International Pediatric Nephrology Association (IPNA) on 7.1.2024

It was inaugurated by the Principal And Controller, SMS Medical College Jaipur, Dr Rajeev Bagarhatta, and Pro Vice-Chancellor Rajasthan University Of Health Sciences, Jaipur, Dr Amarjeet Mehta.

The proceedings began with a brief preview of departmental presentation by the head of the department, Dr.Neha Agarwal especially the recent Sister Renal Centre Collaboration with Great Ormond Street Hospital London. The program focussed on peritoneal dialysis and troubleshooting through a blend of didactic lectures and hands-on workshop. It was well attended by pediatric residents and local paediatricians.

Faculty in attendance were Prof Kjell Tullus (Online Lecture), Prof Amarjeet Mehta, Dr Aditi Sinha, Dr Swati Bhardwaj, Dr Aliza Mittal, Dr Yashu Saini, Dr Ankit Mangla, Dr Alok Upadhyay, Dr Maneesha Bhargava & Dr Vivek Parihar.

The organising committee consisted of Dr Neha Agarwal (Org Secretary), Dr Maneesha Bhargava(Co-org Secretary), Dr Neelam Singh and Dr Piyush Goyal (Workshop Co-ordinators)





Little Survivors

This section highlights the stories of children with kidney disease who have faced their disease boldly and emerged successful.

My Kidney Transplant Journey

'X'16 year old transplant recipient

IKDRC, Ahmedabad



*Dr Kinnari Vala
IKRDC, Ahmedabad*



November 26, 2018, is a day that changed my life forever. It was the day I received a kidney transplant, thanks to my selfless mother who donated one of her kidneys to save my life. Before the transplant, my life was a constant struggle. I was severely underweight, and my low food intake had stalled my growth. At 16, I looked more like a 12-year-old, lacking the strength and energy to keep up with my peers. But all of that changed after the transplant. I began to regain my strength, and my weight started to increase. I grew about 3-4 centimetres in height, and my overall appearance transformed. The hormonal balance restored, and I felt like a new person. However, my journey didn't end there. Prior to the transplant, I had developed bent legs, which affected my mobility and confidence. Post-transplant, I underwent corrective surgery to address this issue. The surgery was a resounding success, renewing my limb strength and replacing my clumsy, knocking knee gait with a smart, energetic, and confident walk. As I regained my health, I discovered new passions and interests. I started playing mobile games, and eventually, I began venturing outdoors to engage in physical activities. My renewed energy also helped me focus on my studies. I completed my 12th commerce with good grades and then cleared my CMA Foundation with excellent marks.

Currently, I'm pursuing my Bachelor of Commerce degree, and I'm in my third year. The kidney transplant and subsequent corrective surgery have brought numerous benefits to my life. I can now enjoy fresh food and fruits, something I couldn't do before. My weight and height are now comparable to those of my peers. Of course, there are still some limitations. I have to be mindful of my physical activities and avoid excessive running. But these minor restrictions pale in comparison to the incredible gains I've made. Throughout this journey, I've received unwavering support from Dr Kinnari Wala, Dr Anshuman Saha, Dr Shahnaz Kapadia, and the entire IKDRC team. Their care, guidance, and encouragement have made an immense difference in my life. In retrospect, my kidney transplant and corrective surgery have been transformative experiences. They've given me a new lease on life, and I'm grateful for every moment.



Clinical Quiz

In this section we describe interesting cases with some important teaching points. In this issue we have used two formats. Do send me emails, which format do you like.

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Dr. SUMANTRA KUMAR RAUT



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An Interesting Case of AKI

Department of Pediatric Nephrology, CMC, Vellore



Dr Srinivasavaradan G



Dr Georgi Mathew

A 3-year-old girl presented with fever, vomiting, and diarrhoea for three days and oliguria for 1 day. There was no history of poor feeding/ lethargy, edema, altered sensorium, or seizures. She was born to a consanguineous parentage and there was a history of early neonatal deaths in her siblings. However, her perinatal period was uneventful. At presentation, she appeared to have some dehydration and had stage 2 hypertension. Systemic examination was unremarkable. Initial laboratory investigations (Table 1) showed severe anemia with evidence of hemolysis (12% schistocytes in peripheral smear), thrombocytopenia, and acute kidney injury (AKI) stage 3. Urine evaluation showed microscopic hematuria with nephrotic range proteinuria. Given hemolytic anemia, and thrombocytopenia with a nephritis-like presentation, the possibility of thrombotic microangiopathy was considered. Stool culture was negative for Shigella and E. coli. She had a normal complement C3 (92 mg/dL; 90-120) and C4 (34 mg/dl; 10-40).



Serology was negative for antinuclear antibodies, hepatitis B, hepatitis C, HIV, scrub typhus, dengue, and typhoid. Homocysteine levels were mildly elevated ($11.5 \mu\text{mol/L}$; 2.6-11) with normal vitamin B12 and folate levels (555 pg/mL and $>20 \text{ ng/mL}$ respectively; 197-771 and 4-23). Antibodies against complement factor H were negative.

1. What are the other possibilities to be considered?
2. Mention common genetic causes associated with TMA



Further evaluation of TMA in our index case revealed a very low ADAMTS13 activity of 2.3% (Hemos IL AcuStar chemiluminescent immunoassay, reference range 60.6 to 130.6 %), and mixing studies showed 73.3%, indicating the absence of inhibitors. Considering the possibility of thrombotic thrombocytopenic purpura (TTP), one unit of cryosupernatant (10 ml/kg) and packed red cells (10 ml/kg) were administered during the hospital stay, along with the management of dehydration. Her serial evaluation showed remission of TMA by day 5 and was discharged on day 6 of illness. Whole exome sequencing showed a homozygous truncating variant (c.4042C>T, p.Gln1348Ter) in exon 29 of ADAMTS13 gene and was classified as likely pathogenic as per ACMG guidelines (PS2, PM3, PP3, PP4), leading to a final diagnosis of Upshaw-Schulman syndrome. At three months of follow-up, she is maintaining normal platelet levels with improvement in haemoglobin and return of creatinine and proteinuria to normal levels.

Discussion

Upshaw-Schulman syndrome (USS) is a rare, autosomal recessive condition caused by the congenital absence or defect of the von Willebrand factor (vWF) cleaving protein, known as ADAMTS13. ADAMTS13 gene (chromosome 9q34) encompasses 29 exons, encoding for a multi-domain zinc containing metalloprotease, a disintegrin-like, thrombospondin type 1 motif, member 13 (ADAMTS13). After thrombosis, ADAMTS13 cleaves vWF and arrests progression. In patients with USS, ultra-large multimers of vWF are not cleaved, leading to the formation of platelet microthrombi in the kidneys, brain, placenta, and heart, leading to renal impairment, neurological symptoms and unfavourable pregnancy outcomes. Over 150 mutations are reported till date, including deletions, insertions, nonsense, missense and, splice-site mutations. Though robust genotype-phenotype correlations are not observed, pre-spacer variants are associated with disease in childhood.

Neonatal and childhood presentation was observed in 38-46% of patients from various ethnicities. Neurologic symptoms dominate the presentation of TTP with headache, transient ischemic attacks, seizures, confusion, and vertigo. Less than 10% of patients reported bleeding though thrombocytopenia is seen in all children. Cardiac involvement is rare and considered to be a bad prognostic sign. Kidney involvement can be seen at initial presentation (25%) as well as later during disease course (10%) and can progress to end stage kidney disease and transplantation in 10%. Post-transplant recurrence of TTP is a cause of concern and can lead to poor outcomes.



Current options of treatment of congenital TTP involves regular prophylactic ADAMTS13 replacement, with fresh frozen plasma, ADAMTS13-containing factor VIII concentrate or rADAMTS13. Significant reduction in the incidence of cerebrovascular thrombosis was observed in patients receiving prophylactic ADAMTS13 replacement. In the UK cohort, 49 (67%) received prophylactic transfusions of either plasma or factor VIII concentrate containing ADAMTS13, with significant reduction in stroke incidence in those receiving prophylaxis (2% vs 17%; $p = .04$). Certain registries have observed good clinical outcomes in up to 30% of patients receiving on-demand ADAMTS13 replacement when compared to prophylactic therapy. The usual frequency of infusions ranges from fortnightly to 3 weekly, with adjustment of infusion frequency according to symptoms and platelet counts. In children with USS in remission, the International Society of Thrombosis and Haemostasis suggests either plasma infusion or wait-and-watch strategy. Congenital TTP resulting from pathogenic variants in ADAMTS13 is rare in children. The wide range of presenting age and clinical manifestations make the diagnosis challenging; and a systematic approach to laboratory evaluation and genetic testing is required for correct diagnosis. Successful management is possible with ADAMTS13 replacement, preventing end organ damage from arterial thromboembolic events.

Table 1: Salient laboratory investigations of the patient

Age of the patient	<i>Current admission</i>					Followup	
	3 year 8 months					1 month	3 month
Haemoglobin (g/dL)	5.6	7	6.8	6.6	7.3	8.3	8.8
Platelets (*103/mm ³)	82	101	89	101	173	1178	389
Reticulocytes (%)		5.34					
Lactate dehydrogenase (U/L)				1456		646	289
Schistocytes (%)	12.2					0.8	0
UP/UC (mg/mg)	3.56					0.12	0.24
Creatinine (mg/dL)	0.97				0.62	0.48	0.36
eGFR (ml/min per 1.73 m ²)	38				60	76	104

eGFR estimated glomerular filtration rate, UP/UC urine protein: creatinine ratio



1. Alwan F, Vendramin C, Liesner R, Clark A, Lester W, Dutt T, et al. Characterization and treatment of congenital thrombotic thrombocytopenic purpura. *Blood*. 2019;133(15):1644–51.
2. Schraner M, Tarasco E, Stalder O, Friedman KD, George JN, Hrachovinova I, et al. Insight into the natural history of hereditary thrombotic thrombocytopenic purpura: short- and long-term outcomes in a longitudinally followed large patient cohort of the international hereditary TTP registry. *Blood*. 2023;142:4003.
3. van Dorland HA, Taleghani MM, Sakai K, Friedman KD, George JN, Hrachovinova I, et al. The International Hereditary Thrombotic Thrombocytopenic Purpura Registry: Key findings at enrolment until 2017. *Haematologica*. 2019;104(10):2107–15.
4. Chrisentery-Singleton T, Boggio LN, Carcao MD, Ibrahimi S, Khan O, Mahajerin A, et al. Long-term follow-up of patients with congenital thrombotic thrombocytopenia purpura receiving a plasma-derived factor VIII (Koate) that contains ADAMTS13. *Haemophilia*. 2023;29(6):1638–45.
5. Mise K, Ubara Y, Matsumoto M, Sumida K, Hiramatsu R, Hasegawa E, et al. Long term follow up of congenital thrombotic thrombocytopenic purpura (Upshaw-Schulman syndrome) on hemodialysis for 19 years: a case report. *BMC Nephrol*. 2013;14(1):156.
6. Sakai K, Fujimura Y, Miyata T, Isonishi A, Kokame K, Matsumoto M. Current prophylactic plasma infusion protocols do not adequately prevent long-term cumulative organ damage in the Japanese congenital thrombotic thrombocytopenic purpura cohort. *Br J Haematol*. 2021;194(2):444–52.
7. Zheng XL, Vesely SK, Cataland SR, Coppo P, Geldziler B, Iorio A, et al. ISTH guidelines for treatment of thrombotic thrombocytopenic purpura. *J Thromb Haemost*. 2020;18(10):2496–502.

Answers to clinical quiz

1. Thrombotic thrombocytopenic purpura - genetic/ acquired, atypical HUS with genetic etiology
2. DGKE, CFH, CFI, CD46, C3, CFB and ADAMTS13



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