

#NephJC Transcript

Healthcare social media transcript of the #NephJC hashtag.

Tue, February 12th 2019, 8:00PM – Tue, February 12th 2019, 11:00PM (America/Toronto).

See #NephJC Influencers/Analytics.



Vasundhara Raghavan @Vasuragh

4 days ago

RT @NephJC: #NephJC in 90 minutes! Paper <https://t.co/vXPPwQPsQR>
@ColumbiaKidney Summary <https://t.co/HRfctGfo1b> @yasar_caliskan @jennie...



Dr Mukesh Rulaniya @RulaniyaDr

4 days ago

RT @edgarvlermamd: Exome sequencing: What clinicians need to know #NephJC
#Nephpearls 🗑️ <https://t.co/o3Fo8NdKiB> <https://t.co/POXHXfK02t>



Divya Bajpai 🇮🇳 @divyaa24

4 days ago

RT @NephJC: #NephJC in 90 minutes! Paper <https://t.co/vXPPwQPsQR>
@ColumbiaKidney Summary <https://t.co/HRfctGfo1b> @yasar_caliskan @jennie...



Allin @alinspring

4 days ago

RT @edgarvlermamd: Diagnostic Utility of Exome Sequencing for Kidney Disease ca.
2019 from @NEJM #VisualAbstract by @divyaa24 #NephJC #Nep...



Matt Sampson @kidneyomicsamps

4 days ago

Few things would preempt me watching a Duke 🏀🐾 basketball game; a #kidneyomics
NephJC is one of them #nephjc



Justin Davis @jbda19

4 days ago

RT @NephJC: #NephJC in 90 minutes! Paper <https://t.co/vXPPwQPsQR>
@ColumbiaKidney Summary <https://t.co/HRfctGfo1b> @yasar_caliskan @jennie...



Anna Burgner MD MEHP @anna_burgner

4 days ago

RT @NephJC: #NephJC in 90 minutes! Paper <https://t.co/vXPPwQPsQR>
@ColumbiaKidney Summary <https://t.co/HRfctGfo1b> @yasar_caliskan @jennie...



Sarah Melville @sarahkmels

4 days ago

RT @edgarvlermamd: Genetic Testing for Chronic Kidney Disease!?! Check out the write up by @rheault_m #NephJC #Nephpearls <https://t.co/d9G...>



Sarah Melville @sarahkmels

4 days ago

RT @edgarvlermamd: The fault lies in our genes!?! Check out the write up by @yasar_caliskan @jenniejlin #NephJC #Nephpearls <https://t.co/ba...>



Nephrology Jrnl Club @NephJC

4 days ago

I am Divya Bajpai @divyaa24, your host for tonight, a nephrologist at KEM hospital Mumbai, #NSMC faculty. I see both adult and pediatric patients and am excited about the applications of this paper to clinical practice #NephJC <https://t.co/TNWAjZsaPQ>



Nephrology Jrnl Club @NephJC

4 days ago

Hi everyone, it's time for #NephJC. Welcome one and all to tonight's discussion about the fascinating topic of exome sequencing in kidney diseases covered in this NEJM paper <https://t.co/6Fzi02iRGE>



Kevin J. Fowler @gratefull080504

4 days ago

I agree @ansakhuja I agree with 100% of @rheault_m comments Well done #nephJC



Edgar V. Lerma 🇵🇭 @edgarvlermamd

4 days ago

Edgar Lerma, Chicago-based ❄️ Nephrologist #NephJC ✈️ On a delayed flight to Miami ☀️ <https://t.co/K9YGlonICI>



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

Swapnil Hiremath, nephrologist from @ottawarenal aka #Snowttawa #NephJC Here today to learn <https://t.co/9mXfw2Fu0g>



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: Hi everyone, it's time for #NephJC. Welcome one and all to tonight's discussion about the fascinating topic of exome sequencing...

Nephrology Jrnl Club @NephJC

4 days ago



Anyone new to #NephJC is always more than welcome. Here's a quick guide on 'How to NephJC.' A particularly warm welcome to new #NSMC interns joining tonight #NephJC <https://t.co/sZIPzPeJkw>

Alex M 🇲🇪 @NephroGuy

4 days ago



Hello! Alex from Mexico. No COI. Mostly lurking tonight. #NephJC

Nephrology Jrnl Club @NephJC

4 days ago



Please introduce yourself (even if you're lurking) and declare any COIs. Don't forget the hashtag. #NephJC

Sarah Melville @sarahkmels

4 days ago



RT @NephJC: Anyone new to #NephJC is always more than welcome. Here's a quick guide on 'How to NephJC.' A particularly warm welcome to new...

Matt Sparks @Nephro_Sparks

4 days ago



Matt Sparks #Pod ⚡ reporting for duty #NephJC

Matt Sampson @kidneyomicsamps

4 days ago



Matt Sampson, peds nephrologist from University of Michigan #nephjc

Diana Mina @DiMiRenalMD

4 days ago



Hi all! Diana here, nephrologist in Dallas. Excited to discuss this topic! Hi @divyaa24 and all my other #NSMC crew! #NephJC

Timothy Yau @Maximal_Change

4 days ago



@hswapnil @OttawaRenal @rolandbastphoto hey swap! looking cold up there #nephJC

Khaled Shawwa @khaledshawwa

4 days ago



Khaled Shawwa, Nephrology Fellow, Rochester, MN. No COI #nephjc

Swapnil Hiremath, MD, MPH @hswapnil

4 days ago



Hope first author from @ColumbiaKidney can join us tonight #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

hi Sarah, Timothy, Matt, Diana welcome to #nephjc

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: Anyone new to #NephJC is always more than welcome. Here's a quick guide on 'How to NephJC.' A particularly warm welcome to new...

**Nephrology Jrnl Club @NephJC**

4 days ago

Check out this great summary by @yasar_caliskan and @jenniejlin <https://t.co/HRfctFXN9D> and a nice commentary by <https://t.co/RZO743V946> by @rheault_m #NephJC**Diana Mina @DiMiRenalMD**

4 days ago

RT @NephJC: Anyone new to #NephJC is always more than welcome. Here's a quick guide on 'How to NephJC.' A particularly warm welcome to new...

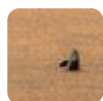
**Edgar V. Lerma 🇵🇭 @edgarvlermamd**

4 days ago

Diagnostic Utility of Exome Sequencing for Kidney Disease ca. 2019 from @NEJM #VisualAbstract by @divyaa24 #NephJC #Nephpearls 👉 <https://t.co/XYvox2PNKg> <https://t.co/xzqu4hl4Ex>**Cathy Quinlan @KidneyCathy**

4 days ago

#nephjc

**Susan Thomas @kidsbeansdoc**

4 days ago

@NephJC Hello. Susan Thomas, Michigan, no COI #nephjc

**Kevin J. Fowler @gratefull080504**

4 days ago

#nephjc Checking in from snowy and cold Chicago tonight COI: Protalix, Retrophin, Hansa Biopharma, Otsuka, Bayer, Gilead, CareDx

**Mya Htwe Nge @mhtwenge**

4 days ago

Hi, Mya from Myanmar 🇲🇲, no COI #nephjc Learning exon... <https://t.co/dHEXniKxHN>

**Bhavnish (Buck MD) @Buck1486**

4 days ago

Hi all, Buck here from OSUWMC. Lurking tonight. #nephjc @NSMCInternship

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

@kidneyomicsamps Hey Matt - glad you could join us tonight! #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

hi cathy, susan #nephjc

**Gary Singer @GarySingerMD**

4 days ago

Gary Singer community nephrologist from STL looking forward to learning about nephrogenomics - no COI #NephJC <https://t.co/EMEuKVBoIB>**L. Parker Gregg @LParkerGregg1**

4 days ago

Parker Gregg, nephrologist in Dallas. No COI. #NephJC #NSMC intern

**Kidney Rebel @kidneyrebel**

4 days ago

ADPKD patient. Exciting paper. Thank you for hosting @divyaa24 Mostly lurking tonight. #NephJC

**Sarah Melville @sarahkmels**

4 days ago

Hi, Sarah Melville, clinical research associate in Atlantic Canada @HeartResearchNB no COI - #NSMC #nephjc

**Anna Burgner MD MEHP @anna_burgner**

4 days ago

Hi all! Ann from @VUMCKidney ere! No coi! #nephjc

**Mario Funes MD @MarioFunesMD**

4 days ago

Mario Funes, PGY2 IM resident in New Brunswick, NJ. #NSMC class 2019. No COI. #NephJC

**Timothy Yau @Maximal_Change**

4 days ago

Tim Yau, @WUNephrology, no COI. Pulled in @WUSTLmed M4 @ccxjwang who is on a medJC sampler tour with me as part of a #SoMe elective. Her first #NephJC

**Cathy Quinlan @KidneyCathy**

4 days ago

Cathy Quinlan, paed neph Melbourne, COI supervising a similar project with @AusGenomics and @MelbGenomics and @kidgenaustralia #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

The visual abstract is by me @divyaa24 #NephJC <https://t.co/YxG1OjsAKk>

**Columbia Nephrology @ColumbiaKidney**

4 days ago

@hswapnil I'm here! #nephjc

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: Check out this great summary by @yasar_caliskan and @jenniejlin <https://t.co/HRfctFXN9D> and a nice commentary by <https://t.co/R...>

**Samira Farouk, MD, MS @ssfarouk**

4 days ago

Samira Farouk, transplant nephrologist @ISMMSKidney. no COI #NephJC

**J. Brian Byrd, MD, MS 🐦 @thebyrdlab**

4 days ago

Brian Byrd, cardiologist and hypertension researcher at University of Michigan. I will be lurking. #nephJC

**Cynthia Wang @ccxjwang**

4 days ago

Hi everyone! This is Cynthia Wang, a 4th year medical student @wustlmed, participating in #nephJC for the first time as part of a #SoMe project mentored by @Maximal_Change. Looking forward to discussing this exciting paper on NGS coming from my undergrad alma mater! #NephJC

**Jennie Lin, MD MTR @jenniejlin**

4 days ago

Hola from the Windy City! Exciting topic and paper to discuss tonight. No COI. #NephJC

**Kevin J. Fowler @gratefull080504**

4 days ago

@hswapnil @ColumbiaKidney I invited him but he had a conflict some of his @ColumbiaKidney colleagues may be joining tonight #nephJC

**Syed Husain @SAHusainMD**

4 days ago

syed ali husain, transplant nephrologist at @ColumbiaKidney. mostly lurking. COI-authors are from my institution #nephjc

**Chi Chu @cdchu**

4 days ago

Hi all! Chi Chu, nephrology fellow, no COI #nephjc

**Diana Mina @DiMiRenalMD**

4 days ago

@Maximal_Change @WUNephrology @WUSTLmed @ccxjwang Wow a #SoMe elective, sounds so cool! #NephJC

**Timothy Yau @Maximal_Change**

4 days ago

@DiMiRenalMD @divyaa24 Bummed to miss KidneyCON this year, have fun w/o me if that's possible #NephJC

**Marvin Gonzalez @MarvinGonzlez16**

4 days ago

Marvin Gonzalez from Nicaragua. No COI..... #NephJC #NSMC

**Nephrology Jrnl Club @NephJC**

4 days ago

@ColumbiaKidney @hswapnil Welcome, Emily! #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: The visual abstract is by me @divyaa24 #NephJC
<https://t.co/YxG1OjsAKk>

**Mario Funes MD @MarioFunesMD**

4 days ago

@ccxjwang @WUSTLmed @Maximal_Change Welcome! It is nice to have you here. #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

Routine housekeeping out of the way first: We recommend you use native client in browser <https://t.co/1SZrrvB3Yf> Tweetdeck. Tchat will truncate tweets at 140 #NephJC

**Kevin J. Fowler @gratefull080504**

4 days ago

@Nephro_Sparks Hi @Nephro_Sparks ! #nephJC

**Anna Burgner MD MEHP @anna_burgner**

4 days ago

Struggling typing on cell phone tonight! It's Anna and here! #NephJC

**Angel Cesar Ortiz @AngelCesarOrtiz**

4 days ago

Hi all. Here Angel Ortiz, last days as nephrology fellow from Mexico @nefrocmnr. #NSMC advisor Excellent topic tonight in #NephJC No COI

**Nephrology Jrnl Club @NephJC**

4 days ago

welcome everyone from @ColumbiaKidney #nephjc

**Arshad Ali @docarshadali**

4 days ago

This is Arshad Ali. Clinical Nephrologist. Michigan. No COI. #NephJC . Ready to learn.

**Syed Husain @SAHusainMD**

4 days ago

@ColumbiaKidney @hswapnil awesome! excited to have you in the discussion, Emily #nephjc

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

First author Emily Groopman is in the house! #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

@ccxjwang @WUSTLmed @Maximal_Change Welcome to #NephJC!

**Nephrology Jrnl Club @NephJC**

4 days ago

Abbreviations for tonight NGS = Next generation sequencing WES = whole exome sequencing WGS = whole genome sequencing #NephJC

**Diana Mina @DiMiRenalMD**

4 days ago

RT @hswapnil: First author Emily Groopman is in the house! #NephJC
<https://t.co/NK0912InfT>**Nephrology Jrnl Club @NephJC**

4 days ago

Welcome Emily #nephjc

**Michelle Starr @mcstarr1**

4 days ago

Hi, Michelle Starr - peds nephrology fellow in Seattle. No COI - excited for this one!
#nephjc

**Vanessa Lerma @VanessaLerma17**

4 days ago

Vanessa Lerma , Nephrology resident in Mexico, #NSMC2019 #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: Abbreviations for tonight NGS = Next generation sequencing WES = whole exome sequencing WGS = whole genome sequencing #NephJC

**Diana Mina @DiMiRenalMD**

4 days ago

RT @NephJC: Abbreviations for tonight NGS = Next generation sequencing WES = whole exome sequencing WGS = whole genome sequencing #NephJC

**Aakash Shingada @aakashshingada**

4 days ago

#NephJC hi. Aakash from New Delhi. Lurking from my on call bed.. Probably the penultimate call duty as a resident.

**Eric Au @ericau**

4 days ago

hi Eric Au, nephrologist from Sydney. #NSMC intern. no COI #nephjc

**Timothy Yau @Maximal_Change**

4 days ago

@gratefull080504 Hi Kevin, hope you're staying warm up there! #NephJC

**Mo Alzubaidi @NephrosisMo**

4 days ago

Mo Alzubaidi form Columbia SC. Lurking tonight #NephJC <https://t.co/D1aaF3Dn1g>

**Sarah Melville @sarahkmels**

4 days ago

RT @NephJC: The visual abstract is by me @divyaa24 #NephJC
<https://t.co/YxG1OjsAKk>

**Columbia Nephrology @ColumbiaKidney**

4 days ago

@hswapnil Excited to join! Look forward to discussing :) #nephjc



Beje Thomas @bthomas215
Beje Thomas no COI just lurking tonight #nephjc

4 days ago



Silvi Shah, MD, MS @silvishah
Hi this is Silvi Shah from UCincy #nephJC

4 days ago



Nephrology Jrnl Club @NephJC
T0a: If NGS is a new term for you like it was for me before reading this paper, just check out this amazing video <https://t.co/JWVhc87c08> #NephJC

4 days ago



Beje Thomas @bthomas215
Great VA #nephjc

4 days ago



Joel Topf, MD FACP @kidney_boy
#NephJC @kidney_boy and @ChangUCanSpare checking in from @TwoJamesSpirits

4 days ago



Arshad Ali @docarshadali
RT @NephJC: Abbreviations for tonight NGS = Next generation sequencing WES = whole exome sequencing WGS = whole genome sequencing #NephJC

4 days ago



Nephrology Jrnl Club @NephJC
@bthomas215 thanks Beje #nephjc

4 days ago



Kevin J. Fowler @gratefull080504
@Maximal_Change Hi @Maximal_Change Trying my best #nephJC

4 days ago




Aakash Shingada @aakashshingada
RT @NephJC: T0a: If NGS is a new term for you like it was for me before reading this paper, just check out this amazing video <https://t.co/...>


4 days ago





Joel Topf, MD FACP @kidney_boy
#NephJC @kidney_boy and @ChangUCanSpare checking in from @TwoJamesSpirits <https://t.co/z3KchCCaxs>

4 days ago


 **Nephrology Jrnl Club @NephJC** 4 days ago
T0a: Also check out this great write up for #nephmadness 2015 on 'GWAS in Nephrology vs Next-Generation Sequencing' <https://t.co/yITbTjXNI> #NephJC


 **Anna Strasma @AStrasma** 4 days ago
Hi, I'm Anna, an IM resident at Baylor! No COI. Excited for #nephjc


 **Marvin Gonzalez @MarvinGonzlez16** 4 days ago
[@hswapnil](#) Wellcome Emily Groopman. #NephJC


 **Itunu @itunuowoyemi** 4 days ago
Hi all, Itunu here transplant fellow [@PITTRenal](#) [@NSMCInternship](#) No COI, glad to be here tonight remember to include #nephjc :)

 **Timothy Yau @Maximal_Change** 4 days ago
[@kidney_boy](#) [@ChangUCanSpare](#) [@TwoJamesSpirits](#) What'cha drinking? #NephJC

 **Swapnil Hiremath, MD, MPH @hswapnil** 4 days ago
[@ColumbiaKidney](#) And BTW, fabulous to see you as a first author on an [@NEJM](#) paper. Nature, nurture & hard work we presume ☐ Hope you are pursuing #nephrology! #NephJC

 **Nephrology Jrnl Club @NephJC** 4 days ago
T0b: Targeted capture & sequencing of the protein-coding regions of the genome through exome sequencing can improve diagnosis & treatment in childhood neurodevelopmental disorders. <https://t.co/uUU1t2t8zz> & <https://t.co/YicoHzc16a> #NephJC

 **Timothy Yau @Maximal_Change** 4 days ago
[@AStrasma](#) Welcome!!!! Glad you could join #NephJC

 **Columbia Nephrology @ColumbiaKidney** 4 days ago
[@NephJC](#) Ha - amazing title! But prefer to see NGS and GWAS as two complimentary vs competing methods for genetic discovery... #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: T0a: If NGS is a new term for you like it was for me before reading this paper, just check out this amazing video <https://t.co/...>



Nephrology Jrnl Club @NephJC

4 days ago

T0c: Exome sequencing has also helped in identifying causal mutations in cancers where genomic diagnosis is routinely used. <https://t.co/JvQiw4tZob> & <https://t.co/A9DQ5XHkHn> #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

#nephjc It is great to see so many new people tonight #nephJC



Gautam Phadke @gautamphadke

4 days ago

gautam phadke nephrologist from the prarie land North Dakota #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@AStrasma welcome to #NephJC Anna!



Nephrology Jrnl Club @NephJC

4 days ago

T0d: Mendelian disorders are a leading cause of CKD in children and account for upto 10% in adults. As patients are asymptomatic till advanced stages it becomes difficult to determine the etiology using standard diagnostic procedures & get labeled as CKD of unknown cause #NephJC



Cathy Quinlan @KidneyCathy

4 days ago

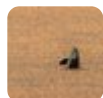
RT @NephJC: T0a: If NGS is a new term for you like it was for me before reading this paper, just check out this amazing video <https://t.co/...>



Kushani @kushjaya

4 days ago

Hi there, Kush Jayasinghe PhD student from Melbourne #nephjc



Susan Thomas @kidsbeansdoc

4 days ago

RT @NephJC: T0c: Exome sequencing has also helped in identifying causal mutations in cancers where genomic diagnosis is routinely used. [htt...](https://t.co/...)



Krzysztof Kiryluk @kirylukk

4 days ago

This is a great visual abstract... @kirylukk checking in; glad to hear Emily could make it @ColumbiaKidney #NephJC



Vanessa Lerma @VanessaLerma17

4 days ago

RT @NephJC: T0c: Exome sequencing has also helped in identifying causal mutations in cancers where genomic diagnosis is routinely used. htt...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@kushjaya hey - welcome to #NephJC look forward to your comments



Jung Hoon Son, M.D. @junghoon_sonMD

4 days ago

Hi #nephjc - Jung Hoon Son, COI - work for a kidney disease focused healthcare startup @pulse_data and coauthored with Krzysztof Kiryluk while at Columbia



ankur gupta @ankurparth

4 days ago

Hi. This is Ankur. Nephrology fellow at The Ottawa Hospital. Trying to learn on twitter from my staff Swapnil. No COI.#NephJC



Susan Thomas @kidsbeansdoc

4 days ago

RT @NephJC: T0d: Mendelian disorders are a leading cause of CKD in children and account for upto 10% in adults. As patients are asymptomatic...



Nephrology Jrnl Club @NephJC

4 days ago

T0e: What percentage of patients with ESKD in your practice have 'unknown' etiology? This number was upto 16% in our practice in India in 2012, likely to have increased now. <https://t.co/VwBbL5mVq6> #NephJC



Vanessa Lerma @VanessaLerma17

4 days ago

RT @NephJC: T0d: Mendelian disorders are a leading cause of CKD in children and account for upto 10% in adults. As patients are asymptomatic...



Dr. Michelle Gumz @MLGumz

4 days ago

RT @KidneyCathy: Fantastic summary by @rheault_m in advance of #NephJC <https://t.co/5VpsJ9yX05>



Matt Sampson @kidneyomicsamps

4 days ago

RT @KidneyCathy: Fantastic summary by @rheault_m in advance of #NephJC
<https://t.co/5VpsJ9yX05>



Charlie Hall @CharlesHallMD

4 days ago

Hi, I'm Charlie, Renal SHO from London and #nsmc intern. Working a night shift tonight so mainly lurking. #nephjc



Nephrology Jrnl Club @NephJC

4 days ago

@kirylukk @ColumbiaKidney Thanks @kirylukk glad u liked the VA #nephjc



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0a: If NGS is a new term for you like it was for me before reading this paper, just check out this amazing video <https://t.co/...>



Eric Au @ericau

4 days ago

RT @NephJC: #NephJC in 90 minutes! Paper <https://t.co/vXPPwQPsQR>
@ColumbiaKidney Summary <https://t.co/HRfctGfo1b> @yasar_caliskan @jennie...



Vighnesh Walavalkar @vighnesh_w

4 days ago

@NephJC Hi, Vighnesh from San Francisco, CA. No COI. #NephJC



OLSUKIDNEY @olsukidney

4 days ago

#kidneysu representing LSU Nephrology SHREVEPORT now Oschner LSU Partner Hospital #NephJC



Matt Sparks @Nephro_Sparks

4 days ago

What a crowd on #nephjc tonight ! Join us



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@kirylukk @ColumbiaKidney welcome! another author/expert in the house tonight at #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0a: Also check out this great write up for #nephmadness 2015 on 'GWAS in Nephrology vs Next-Generation Sequencing' <https://t.c...>



Jennie Lin, MD MTR @jenniejlin

4 days ago

@Nephro_Sparks Hot and very relevant topic in the era of precision medicine #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@junghoon_sonMD @pulse_data thanks for joining tonight #NephJC



Samira Farouk, MD, MS @ssfarouk

4 days ago

@NephJC T0e: When we do initial evaluations for potential #kidneytransplant recipients...some days feels like all of them #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

T0f: Smaller studies support the usefulness of exome sequencing for the diagnosis of early onset nephropathy. <https://t.co/CkQLdGcO10> #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0b: Targeted capture & sequencing of the protein-coding regions of the genome through exome sequencing can improve diagnosis &...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: T0e: What percentage of patients with ESKD in your practice have 'unknown' etiology? This number was upto 16% in our practice i...



Sayna Norouzi @SaynaNorouzi

4 days ago

Hi! This is Sayna; nephrology fellow. No Col. #nephjc #NSMC



Mya Htwe Nge @mhtwenge

4 days ago

@NephJC #nephJC ~20% have unknown cause



Gary Singer @GarySingerMD

4 days ago

@NephJC Suspect as many as 15-20% since many present late to neph with CKD stage 4 and HTN and biopsy unlikely to yield Dx #NephJC



Paul A Welling @PAWellingMD

4 days ago

arriving late for discussion of this cool study, no COI #nephjc



Timothy Yau @Maximal_Change

4 days ago

@NephJC Depends a lot on the confidence of the clinician labeling something as "hypertensive nephrosclerosis" or other such vague terms. But I'd say a good 20% we don't have enough confidence to give a strong etiology #NephJC



Cathy Quinlan @KidneyCathy

4 days ago

@NephJC I think in Australia it is around 6% - but unsure about paediatrics #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@NephJC lots often gets labelled as 'ischemic nephropathy' or 'non proteinuric CKD' or some basket diagnosis #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC htExomtp://ow.ly/w6FM50kefzL hope She can join us tonight <https://t.co/IUUcxDorlh>



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0c: Exome sequencing has also helped in identifying causal mutations in cancers where genomic diagnosis is routinely used. htt...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...



Arunkumar @Arunkr_Dr

4 days ago

Hello!! Arunkumar from Delhi.. no COI.. #NephJC



Pablo Garcia @PabloGarciaMD

4 days ago

Hi this is Pablo Garcia, first year nephrology fellow. No Col. #nephjc



Nephrology Jrnl Club @NephJC

4 days ago

@KidneyCathy Wow 6% is great . U are doing a very good job at diagnosing #NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@KidneyCathy @NephJC 6% is not bad! But of course we want 0. #NephJC



Timothy Yau @Maximal_Change

4 days ago

@hswapnil @NephJC I don't do this myself, but if I ever see something that says "chronic glomerulonephritis" it is code for "I have no clue." #NephJC



L. Parker Gregg @LParkerGregg1

4 days ago

@NephJC I feel like we would have more unclear cause diagnoses if we were more aggressive about doing kidney biopsies...lots of presumed 2/2 DM and HTN #NephJC



Itunu @itunuwoyemi

4 days ago

I could not agree more. Thanks to care everywhere and EHR search buttons sometimes it's easier to just search for 'biopsy' lucky only a few times #nephjc



Samira Farouk, MD, MS @ssfarouk

4 days ago

@KidneyCathy @NephJC What % do you biopsy?! 6% is incredible. #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

Hope this will build upon the momentum of the @KPMProject + accelerate early diagnosis of #kidneydiseases + early intervention for #kidneydiseases #nephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0e: What percentage of patients with ESKD in your practice have 'unknown' etiology? This number was upto 16% in our practice i...



Alex M \$□□ @NephroGuy

4 days ago

RT @hswapnil: First author Emily Groopman is in the house! #NephJC
<https://t.co/NK0912InfT>

**Eric Au @ericau**

4 days ago

@ssfarouk @NephJC Wonder if we should be using genetic testing more often in these patients as part of their transplant workup #nephjc

**Mo Alzubaidi @NephrosisMo**

4 days ago

@NephJC 20ish% #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @LParkerGregg1: @NephJC I feel like we would have more unclear cause diagnoses if we were more aggressive about doing kidney biopsies.....

**Timothy Yau @Maximal_Change**

4 days ago

@PabloGarciaMD Hey Pablo, hope all is well with you! #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

T0h: The role of exome sequencing in broader ESRD adult population, where most of the cases are sporadic, was unknown until today's NEJM paper came out #NephJC

**Michelle Starr @mcstarr1**

4 days ago

@NephJC Maybe about 10% of our peds patients? We have a large group of anatomy abnormalities (eg valves) but then a lot that we don't really know why they have ESKD... #nephjc

**Neeraj Inamdar @nainamdar**

4 days ago

RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...

**Matt Sparks @Nephro_Sparks**

4 days ago

@NephJC Of course some of the diabetes could also have underlying genetic component #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

@gratefull080504 @KPMProject Need such projects here in India/Asia #NephJC



L. Parker Gregg @LParkerGregg1

4 days ago

@Nephro_Sparks @NephJC The more people who have diabetes, the more people who will have diabetes and also something else #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@ericau @ssfouk @NephJC I think that is almost becoming mandatory, particularly if living related donors are being considered #nephjc



Mo Alzubaidi @NephrosisMo

4 days ago

RT @Nephro_Sparks: What a crowd on #nephjc tonight ! Join us



Jennie Lin, MD MTR @jenniejlin

4 days ago

@LParkerGregg1 @NephJC Yes, and I disagree with people who biopsy only if they suspect GN to use biopsy to confirm what they had already suspected. But maybe genetics can be a common ground since no bleeding risk. #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

If genetic testing is being done in #kidneytransplant why hasn't it been done earlier? #nephJC



Timothy Yau @Maximal_Change

4 days ago

@NephJC @rheault_m @CJASN As pointed out in her commentary, pretty huge treatment implications, since I wouldn't be throwing prednisone at these FSGS patients if I knew they had a mutation #NephJC



Yasar Caliskan @yasar_caliskan

4 days ago

I am sure most of the diagnosis of chronic glomerulonephritis or HT nephrosclerosis or diabetic nephropathy have no biopsy confirmation. #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @ssfouk: @NephJC T0e: When we do initial evaluations for potential #kidneytransplant recipients...some days feels like all of them #Ne...



Larissa Krüger @nephron_andon

4 days ago

Larissa here from UConn, no COI. Joining a little late, but looking forward to discussion! #NephJC @NSMCInternship

**Jennie Lin, MD MTR** @jenniejlin

4 days ago

@Nephro_Sparks @NephJC And enter @ChristosArgyrop and @KSusztak #NephJC

**Sarah Melville** @sarahkmels

4 days ago

RT @NephJC: T0f: Smaller studies support the usefulness of exome sequencing for the diagnosis of early onset nephropathy. <https://t.co/CkQL...>**Matt Sparks** @Nephro_Sparks

4 days ago

Finding novel genetic mutations and labeling as causative is still a serious business and needs to be done meticulously #NephJC

**Matt Sampson** @kidneyomicsamps

4 days ago

@jenniejlin @LParkerGregg1 @NephJC I guess if you are collecting DNA via saliva 😊 #nephjc

**Swapnil Hiremath, MD, MPH** @hswapnil

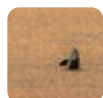
4 days ago

@jenniejlin @LParkerGregg1 @NephJC biopsy for diagnosis when no potential treatment is not easy just saying #nephJC

**Nephrology Jrnl Club** @NephJC

4 days ago

Amazing discussion so far, and now...onto the paper #NephJC.

**Susan Thomas** @kidsbeansdoc

4 days ago

@kidneyomicsamps @ericau @ssfouk @NephJC Important in determining risk of recurrence, and risk to donor. #nephjc

**Gary Singer** @GarySingerMD

4 days ago

@LParkerGregg1 @Nephro_Sparks @NephJC As many as 20% of diabetics with proteinuria will have another Dx on Bx #NephJC

**Adrián Chávez** @ChavinhoMD

4 days ago

RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...

**Matt Sparks @Nephro_Sparks**

4 days ago

@jenniejlin @NephJC @ChristosArgyrop @KSusztak Just saying the we bucket as DM too often #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

@jenniejlin @LParkerGregg1 Yes It is less invasive, but still out of reach of most of our patients #noMoney #NephJC

**Kidney Rebel @kidneyrebel**

4 days ago

About how much does it cost to exome sequence a patient? How quickly is the cost coming down? #NephJC.

**Tiffany Caza @Tiff_Caza**

4 days ago

@NephJC @rheault_m @CJASN Is Alports under recognized? Seems like a high percentage. #NephJC Great VA @rheault_m

**L. Parker Gregg @LParkerGregg1**

4 days ago

@hswapnil @jenniejlin @NephJC For now...but once we hear more from @KPMProject maybe that will change #NephJC

**Jennie Lin, MD MTR @jenniejlin**

4 days ago

@hswapnil @LParkerGregg1 @NephJC but as @rheault_m had said in her great commentary, every patient deserves a name for his/her disease. Even if no treatment. #NephJC

**Gates Colbert, MD @DoctorGates**

4 days ago

Dallas Nephrologists. Joining late. It's almost Nephmadness season. 🍷 #NephJC

**Matt Sampson @kidneyomicsamps**

4 days ago

@Maximal_Change @NephJC @rheault_m @CJASN A proportion of NS patients with Mendelian pathogenic variants in fact respond to prednisone. Would not let a genetic diagnosis preclude me from at least trying steroids #nephjc

Nephrology Jrnl Club @NephJC

4 days ago



Methods: T1A: Proband only exome sequencing was performed on 3315 pts from 2 independent cohorts. First 1128 were from AURORA which included patients aged 50-80 and all-cause ESKD on HD >3 months. #NephJC <https://t.co/3tG5sISY8F>

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

@Tiff_Caza @NephJC @rheault_m @CJASN Alport, not Alport's :-) #NephJC

**Kevin J. Fowler @gratefull080504**

4 days ago

@Nephro_Sparks : Great point need to move away from #CKD to #kidneydiseases It is not a monolithic disease Some patients are blamed for their #dkd when #genetics have a role #nephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @kidneyomicsamps: @Maximal_Change @NephJC @rheault_m @CJASN A proportion of NS patients with Mendelian pathogenic variants in fact respo...

**Matt Sampson @kidneyomicsamps**

4 days ago

Could not agree more #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

T1B: Remaining 2187 were from CUMC genetic study of CKD cohort which included patients with renal failure requiring dialysis or transplantation, serum creatinine > 1.5 mg/dL [>1.3 in women] or the presence of severe proteinuria or hematuria #NephJC

**Justin Davis @jbda19**

4 days ago

@NephJC Hi, Justin from the #NSMC internship. Sorry I'm a little late, busy day at work here in Aus. I'll be watching with curiosity #Nephjc

**Cathy Quinlan @KidneyCathy**

4 days ago

@Nephro_Sparks Absolutely - important to be able to look at the phenotype and make sure the genotype matches - eg 2 pathogenic HOGA mutations in a patient with cystic kidney disease. Prospective, real life approach. #nephjc



Timothy Yau @Maximal_Change

4 days ago

@GarySingerMD @LParkerGregg1 @Nephro_Sparks @NephJC I was taught up to a third. Type 1s a little different, I think their timeline is more predictable #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...



Nephrology Jrnl Club @NephJC

4 days ago

T1C: Genomic DNA was isolated from patient samples and sequenced at Columbia Institute for genomic medicine. To facilitate identification of pathogenic variants a manually curated list of 625 genes associated with Mendelian forms of genitourinary diseases was used #NephJC



Marvin Gonzalez @MarvinGonzlez16

4 days ago

@NephJC In my clinical practice: 25-30% of my patients have CKDu, but the prevalence is higher in the lowland compared to the highland. #NephJC #NSMC2019



Yasar Caliskan @yasar_caliskan

4 days ago

Sorry not to confirm just to diagnose GN biopsy is needed. There is no clinical diagnosis of GN as far as I know, #NephJC



Ankit Sakhuja, MBBS FACP FASN @ansakhuja

4 days ago

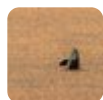
Ankit Sakhuja, no coi #nephJC #NSMC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@NephJC what made the authors combine two different cohorts? Why not just CUMC @Columbiakidney #NephJC



Susan Thomas @kidsbeansdoc

4 days ago

RT @kidneyomicsamps: @Maximal_Change @NephJC @rheault_m @CJASN A proportion of NS patients with Mendelian pathogenic variants in fact respo...

**Michelle Starr** @mcstarr1

4 days ago

@kidneyomicsamps @Maximal_Change @NephJC @rheault_m @CJASN So should we use treatment responsiveness in order to determine when to test? What's your approach? #nephjc

**Eric Au** @ericau

4 days ago

@gratefull080504 That's a good point - perhaps if genetic testing is more widely available it would be done earlier as part of the workup of their initial kidney disease #nephjc

**Nephrology Jrnl Club** @NephJC

4 days ago

T1D: Diagnostic variants were those that were classified as "pathogenic" or "likely pathogenic" according to the American College of Medical Genetics and Genomics (ACMG) guidelines & were explicative of the patient's nephropathy #NephJC <https://t.co/hb3U2jjg5D>

**Kevin J. Fowler** @gratefull080504

4 days ago

@jenniejlin : Without a doubt Starting identifying a #kidneydisease + possibly treatments will be developed once the disease is understood #nephJC

**Neeraj Inamdar** @nainamdar

4 days ago

RT @Maximal_Change: @NephJC @rheault_m @CJASN As pointed out in her commentary, pretty huge treatment implications, since I wouldn't be thr...

**Michelle Starr** @mcstarr1

4 days ago

@kidneyomicsamps @Maximal_Change @NephJC @rheault_m @CJASN Or should we be giving them all CoQ10? #nephjc

**Krzysztof Kiryluk** @kirylukk

4 days ago

Very good point about living donors. Many of the patients included in the paper reached ESRD and received a kidney transplant from a living donor... #nephjc

**Nephrology Jrnl Club** @NephJC

4 days ago

T1E: Also, CUMC patients' sequence data was separately analyzed for pathogenic variants in the 59 genes that would be recommended by the ACMG for reporting as medically actionable secondary findings to patients undergoing genomic sequencing #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @gratefull080504: Hope this will build upon the momentum of the @KPMProject + accelerate early diagnosis of #kidneydiseases + early inte...



Jennie Lin, MD MTR @jenniejlin

4 days ago

@KSusztak @Nephro_Sparks @NephJC @ChristosArgyrop CC: @hswapnil #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@mcstarr1 @Maximal_Change @NephJC @rheault_m @CJASN It's complicated- everyone should have a chance to remit with steroids, in my opinion, unless CLINICAL, not genetic indication not to. But genetic testing should be done early if there is a concern as it could help dictate treatment in those not clearly SSNS #nephjc



Cathy Quinlan @KidneyCathy

4 days ago

RT @mcstarr1: @kidneyomicsamps @Maximal_Change @NephJC @rheault_m @CJASN Or should we be giving them all CoQ10? #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@mcstarr1 @kidneyomicsamps @Maximal_Change @NephJC @rheault_m @CJASN coenzyme Q10? expand - maybe I am missing something #NephJC



Khaled Shawwa @khaledshawwa

4 days ago

@hswapnil @jenniejlin @LParkerGregg1 @NephJC Agree. Also a test is likely to yield results that can potentially change management of a patient if there is some level of suspicion (or pretest probability) #nephjc



Nephrology Jrnl Club @NephJC

4 days ago

@mcstarr1 @kidneyomicsamps @Maximal_Change @rheault_m @CJASN This is important. We have also given steroid trial in selected patients with mendelian disorders proven when biopsy is favorable. Has worked #NephJC



Vanessa Lerma @VanessaLerma17

4 days ago

It would be interesting to have that study in babies with mothers with CKD #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@hswapnil @NephJC as in many cases, having multiple independent cohorts gives a deeper understanding of the value of genetic testing, in different contexts #nephjc



Harish Seethapathy @BetterCallSeeth

4 days ago

Hi everyone! This is Harish, Renal Fellow from MGH. Newbie! No Col #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

T1F: Any thoughts about methods? Any concerns? #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @gratefull080504: If genetic testing is being done in #kidneytransplant why hasn't it been done earlier? #nephJC <https://t.co/fATjtClydz>



Yasar Caliskan @yasar_caliskan

4 days ago

Are they living-related kidney tx and any genetic testing before donation for donors #NephJC #kirylukk



Timothy Yau @Maximal_Change

4 days ago

@NephJC One thing that struck me as I read the paper was how important the reference "normal" is. Outside of reported cases, I never understand how they know if a mutation is pathogenic or not #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @ColumbiaKidney: @hswapnil @NephJC as in many cases, having multiple independent cohorts gives a deeper understanding of the value of ge...



Itunu @itunuowoyemi

4 days ago

@hswapnil @NephJC @ColumbiaKidney Can we assume it was to have some sort of control by combining different cohorts @ColumbiaKidney @hswapnil #NephJC ?



Ankit Sakhuja, MBBS FACP FASN @ansakhuja

4 days ago

@SaynaNorouzi Hi Sayna #NephJC



Cathy Quinlan @KidneyCathy

4 days ago

Was there any way to feed back these results to the AURORA cohort? Esp for the AD conditions in patients who may have children or be considering expanding their family or live donation? #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

#NephJC



Cynthia Wang @ccxjwang

4 days ago

@NephJC Wondering what were the main advantages to doing WES vs targeted gene sequencing in this study given that genes of interest in relation to CKD were known and defined? Is it because it's not that much harder / more expensive to do anyway and gives more info? #NephJC



Gary Singer @GarySingerMD

4 days ago

@Maximal_Change @LParkerGregg1 @Nephro_Sparks @NephJC You are correct sir - Should all patients with diabetes have a kidney biopsy? <https://t.co/ehoOxqq2SU> #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@Maximal_Change @NephJC we use the ACMG guidelines to help assess the pathogenicity of mutations <https://t.co/Uxk4xPDoXY> #nephjc



Franklin Loachamin @FranLoachamin

4 days ago

Hi Franklin Loachamin #HCAM #NephJC im too late nephped Quito Ecuador



Matt Sampson @kidneyomicsamps

4 days ago

That's a big deal. Have you published this? Also gets to @Nephro_Sparks point about actually knowing pathogenicity of rare variants #NephJC #kidneyomics



Michelle Starr @mcstarr1

4 days ago

@hswapnil @kidneyomicsamps @Maximal_Change @NephJC @rheault_m @CJASN I've had a few patients where genetic diagnosis in NS lead to treatment with CoQ10 and eventually response #nephjc



Jennie Lin, MD MTR @jenniejlin

4 days ago

@NephJC @ColumbiaKidney did you guys also analyze the rest of the genome, or just the 625 prioritized genes? If so, any novel hits? #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

Now. Moving on to T2 for the results. #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @jenniejlin: @NephJC @ColumbiaKidney did you guys also analyze the rest of the genome, or just the 625 prioritized genes? If so, any nov...



Kevin J. Fowler @gratefull080504

4 days ago

#nephjc We must have 10-15 new voices tonight Great work @hswapnil @kidney_boy @Nephro_Sparks



Timothy Yau @Maximal_Change

4 days ago

@NephJC @mcstarr1 @kidneyomicsamps @rheault_m @CJASN Goes to show that mendelian disease and an alteration in the immune system are not mutually exclusive, and could represent 2-hits #NephJC



Matt Sparks @Nephro_Sparks

4 days ago

@GarySingerMD @Maximal_Change @LParkerGregg1 @NephJC My point was that even though the biopsy findings looked like diabetic kidney disease this could have been enhanced by and mutation driving the phenotype #NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@NephJC @ColumbiaKidney sorry I meant exome! #NephJC



Fitsum Tilahun MD @fitse_t

4 days ago

@NephJC Joining late..but caught up the thread No COI #nephjc



Columbia Nephrology @ColumbiaKidney

4 days ago

@jenniejlin @NephJC we focused on known genes, since for diagnostic analysis you look only at genes known to be assoc w/human disease #nephjc



Syed Husain @SAHusainMD

4 days ago

@KSusztak @Nephro_Sparks @jenniejlin @NephJC @ChristosArgyrop exactly. link: <https://t.co/cXHEccw1av> #nephjc



Matt Sampson @kidneyomicsamps

4 days ago

@mcstarr1 @hswapnil @Maximal_Change @NephJC @rheault_m @CJASN post the link to your cool paper #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

T2A: The cohort was predominantly adult (3037 patients [91.6%] were >21 years of age), and 1179 patients (35.6%) reported having non-European ancestry #NephJC <https://t.co/wqg9f1mCoR>



Columbia Nephrology @ColumbiaKidney

4 days ago

@jenniejlin @NephJC but there are many opportunities for novel gene discovery with our exome dataset! #nephjc



Nephrology Jrnl Club @NephJC

4 days ago

T2B: Cohort represented all major categories of nephropathy, including nephropathy of unknown origin [8.5%]. 64.7% had ESKD. 28.3% had a positive family history of kidney disease #NephJC



Neeraj Inamdar @nainamdar

4 days ago

@ColumbiaKidney @hswapnil @NephJC How much do you think % will increase by adding WGS and considering variants of unknown significance? #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@KidneyCathy tough ethical Q...i think standard (unstaifactory) answer is no, but would be happy to be corrected @ColumbiaKidney #NephJC



Franklin Loachamin @FranLoachamin

4 days ago

RT @kidneyomicsamps: @Maximal_Change @NephJC @rheault_m @CJASN A proportion of NS patients with Mendelian pathogenic variants in fact respo...

Nephrology Jrnl Club @NephJC

4 days ago



T2C: Diagnostic variants were detected in 307 of the 3315 patients (9.3%), encompassing 66 distinct monogenic disorders. Diagnostic yield was highest in patients congenital or cystic renal disease (23.9%) and in patients with “nephropathy of unknown origin” (17.1%) #NephJC <https://t.co/xySTJyYYxr>

**Samaya @TarSamMD**

4 days ago

Sorry everyone late to the party Samaya from Houston no Col #NephJC

**Michelle Starr @mcstarr1**

4 days ago

@kidneyomicsamps @hswapnil @Maximal_Change @NephJC @rheault_m @CJASN Check it out here: <https://t.co/RmErTreKxg> #nephjc

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: T2A: The cohort was predominantly adult (3037 patients [91.6%] were >21 years of age), and 1179 patients (35.6%) reported havin...

**Matt Sparks @Nephro_Sparks**

4 days ago

@SAHusainMD @KSusztak @jenniejlin @NephJC @ChristosArgyrop Still this just buckets based on pathologic phenotype and the putative mutation could push the phenotype in a way that is synonymous with phenotype just worsening and quicker. #NephJC

**Columbia Nephrology @ColumbiaKidney**

4 days ago

@nainamdar @hswapnil @NephJC adding variants of uncertain sig (VUS) - see the appendix 1, ~86% of cases have at least one VUS... #nephjc

**Sarah Melville @sarahkmels**

4 days ago

RT @DoctorGates: Dallas Nephrologists. Joining late. It's almost Nephmadness season. 🍷 #NephJC

**Krzysztof Kiryluk @kirylukk**

4 days ago

Needed to demonstrate generalizability. Columbia is a tertiary referral and might have been enriched for genetic diagnoses. The amazing thing is that these observations seem to replicate in another large independent cohort. #NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@nainamdar @ColumbiaKidney @hswapnil @NephJC Also, tech question since i don't do exomes - are there enough reads into intron/exon boundaries for splice site variants on the intron side? #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @kiryLUkk: Needed to demonstrate generalizability. Columbia is a tertiary referral and might have been enriched for genetic diagnoses. T...



Marvin Gonzalez @MarvinGonzlez16

4 days ago

@kidneyomicsamps @jenniejlin @LParkerGregg1 @NephJC A researcher from Harvard is collection saliva from patients with CKDu, because he wants to perform DNA sequence study. #NSMC2019 #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@ColumbiaKidney @nainamdar @hswapnil @NephJC What percentage of controls have at least one VUS? #nephjc



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: Methods: T1A: Proband only exome sequencing was performed on 3315 pts from 2 independent cohorts. First 1128 were from AURORA...



Nephrology Jrnl Club @NephJC

4 days ago

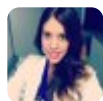
T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients. #NephJC <https://t.co/TezRKwvOSL>



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @mcstarr1: @kidneyomicsamps @hswapnil @Maximal_Change @NephJC @rheault_m @CJASN Check it out here: <https://t.co/RmErTreKxg> #nephjc



Vanessa Lerma @VanessaLerma17

4 days ago

RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....



John Montford, MD @JohnRMontford

4 days ago

#nephjc Hello from Denver, lurker tonight, no coi. Love the study, getting CKDu genetic screening FOMO from this.



Matt Sampson @kidneyomicsamps

4 days ago

@jenniejlin @nainamdar @ColumbiaKidney @hswapnil @NephJC I think yes, at least in our data #nephjc



Sarah Melville @sarahkmels

4 days ago

RT @gratefull080504: @Nephro_Sparks : Great point need to move away from #CKD to #kidneydiseases It is not a monolithic disease Some patien...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....



Arshad Ali @docarshadali

4 days ago

RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....



Matt Sampson @kidneyomicsamps

4 days ago

@MarvinGonzlez16 @jenniejlin @LParkerGregg1 @NephJC What a great idea. Hope it is helpful #nephjc



Timothy Yau @Maximal_Change

4 days ago

@ccxjwang @NephJC My guess is cost factors into it. NGS is cheaper than it used to be, but still pretty expensive for out of pocket. e.g. if insurance doesn't cover it, whole exome sequencing runs around 6K for a nephrotic syndrome panel #NephJC



Mario Funes MD @MarioFunesMD

4 days ago

It would be interesting to see this study done in patients with mesoamerican nephropathy with no conventional risk factors for CKD. #NephJC

Nephrology Jrnl Club @NephJC

4 days ago



T2E: A family history of kidney disease and a clinical diagnosis of congenital or cystic renal disease and nephropathy of unknown origin were independent predictors of having a genetic diagnosis #NephJC <https://t.co/IGvUcU3gQc>

J S Kumar @docchennai

4 days ago



RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....

Franklin Loachamin @FranLoachamin

4 days ago



RT @yasar_caliskan: Sorry not to confirm just to diagnose GN biopsy is needed. There is no clinical diagnosis of GN as far as I know, #Neph...

Jennie Lin, MD MTR @jenniejlin

4 days ago



@MarioFunesMD fructose metabolism variants! #NephJC

Susan Thomas @kidsbeansdoc

4 days ago



RT @MarioFunesMD: It would be interesting to see this study done in patients with mesoamerican nephropathy with no conventional risk factor...

Krzysztof Kiryluk @kirylukk

4 days ago



Agree, still a major clinical challenge. ACMG criteria help, but still not 100%... #NephJC

Timothy Yau @Maximal_Change

4 days ago



RT @kirylukk: Needed to demonstrate generalizability. Columbia is a tertiary referral and might have been enriched for genetic diagnoses. T...

Matt Sampson @kidneyomicsamps

4 days ago



@MarioFunesMD I predict that a GWAS using common alleles would be equally, if not more, high-yield than WES. Seems like a secular change in prevalence, indicating perhaps a new gene-environment interaction #nephjc

Swapnil Hiremath, MD, MPH @hswapnil

4 days ago



RT @NephJC: T2E: A family history of kidney disease and a clinical diagnosis of congenital or cystic renal disease and nephropathy of unkno...



Jennie Lin, MD MTR @jenniejlin

4 days ago

@Maximal_Change @ccxjwang @NephJC Wow, really? someone quoted 800 bucks? but I guess 6K is for CLIA? #NephJC



Timothy Yau @Maximal_Change

4 days ago

@kirylukk brilliant design, thx #NephJC



Franklin Loachamin @FranLoachamin

4 days ago

RT @NephJC: T1D: Diagnostic variants were those that were classified as “pathogenic” or “likely pathogenic” according to the American Colle...



Sarah Melville @sarahkmels

4 days ago

RT @kirylukk: Very good point about living donors. Many of the patients included in the paper reached ESRD and received a kidney transplant...



Nephrology Jrnl Club @NephJC

4 days ago

@MarioFunesMD There was some data presented at #isncon 2018 with NGS of Indian CKDu by SGPGI group. They had also identified families with same defect. Awaiting publication. #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T1D: Diagnostic variants were those that were classified as “pathogenic” or “likely pathogenic” according to the American Colle...



Kevin J. Fowler @gratefull080504

4 days ago

@Maximal_Change : Tim, I think people living with #kidneydisease + patient organizations need to weigh in on this issue. I see NGS as way to disrupt the #dialysis ecosystem which has stifled innovation What about the \$34 billion spent in US on #dialysis #nephJC



Sarah Melville @sarahkmels

4 days ago

RT @GarySingerMD: @Maximal_Change @LParkerGregg1 @Nephro_Sparks @NephJC You are correct sir - Should all patients with diabetes have a kidn...

Nephrology Jrnl Club @NephJC

4 days ago



T2F: In 89% of the CUMC patients with genetic findings from the exome sequencing, the molecular diagnosis had implications to alter clinical management. A subtype of disease was identified in 65 CUMC patients, & disease was reclassified in 18 patients #NephJC <https://t.co/SUmGPeujxd>

Moumita Barua @MoumitaBaruaTO

4 days ago



RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...

Franklin Loachamin @FranLoachamin

4 days ago



RT @NephJC: T2C: Diagnostic variants were detected in 307 of the 3315 patients (9.3%), encompassing 66 distinct monogenic disorders. Diagno...

Mario Funes MD @MarioFunesMD

4 days ago



@NephJC Nice, waiting to see the results. #NephJC

Swapnil Hiremath, MD, MPH @hswapnil

4 days ago



RT @NephJC: T2F: In 89% of the CUMC patients with genetic findings from the exome sequencing, the molecular diagnosis had implications to a...

Columbia Nephrology @ColumbiaKidney

4 days ago



@kidneyomicsamps @nainamdar @hswapnil @NephJC we did not assess internal controls for VUS in this paper. But another paper in the annals gives a good estimate, based on different degrees of stringent filtering... <https://t.co/G4QCRrWWk1> #nephjc

Matt Sampson @kidneyomicsamps

4 days ago



@gratefull080504 @Maximal_Change The more that papers are published like @ColumbiaKidney that show that testing matters (has clinical utility), the more likely insurers will cover it #nephjc

Nephrology Jrnl Club @NephJC

4 days ago



T2G: 62% of patients with COL4A3, COL4A4, or COL4A5 mutations did not have clinical diagnoses of Alport syndrome or thin basement membrane disease. This detection would indicate ophthalmologic and otolaryngologic referral #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @SAHusainMD: @KSusztak @Nephro_Sparks @jenniejlin @NephJC @ChristosArgyrop exactly. link: <https://t.co/cXHEccw1av> #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @ColumbiaKidney: @kidneyomicsamps @nainamdar @hswapnil @NephJC we did not assess internal controls for VUS in this paper. But another pa...



Neeraj Inamdar @nainamdar

4 days ago

RT @ColumbiaKidney: @kidneyomicsamps @nainamdar @hswapnil @NephJC we did not assess internal controls for VUS in this paper. But another pa...



Sarah Melville @sarahkmels

4 days ago

RT @ColumbiaKidney: @jenniejlin @NephJC but there are many opportunities for novel gene discovery with our exome dataset! #nephjc



Susan Thomas @kidsbeansdoc

4 days ago

RT @kidneyomicsamps: @gratefull080504 @Maximal_Change The more that papers are published like @ColumbiaKidney that show that testing matter...



Timothy Yau @Maximal_Change

4 days ago

@jenniejlin @ccxjwang @NephJC Wow, that being said, that was 2.5 years ago, so maybe cheaper now? And it was a unique case of non-insurance coverage. #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@ColumbiaKidney @nainamdar @hswapnil @NephJC So what is your rough guess? #nephjc



Nikhil Shah @dr_nikhilshah

4 days ago

#nephjc Joining in late. Nikhil Shah, Clinical Nephrologist from Edmonton @UAlberta @UAlbertaNeph, no COI!



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @kidneyomicsamps: @gratefull080504 @Maximal_Change The more that papers are published like @ColumbiaKidney that show that testing matter...

**Nephrology Jrnl Club @NephJC**

4 days ago

T2H: In 15 patients (16%) with a clinical diagnosis of focal segmental glomerulosclerosis, the genetic diagnosis could disfavor unnecessary immunosuppressive therapy #NephJC

**Franklin Loachamin @FranLoachamin**

4 days ago

RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....

**Timothy Yau @Maximal_Change**

4 days ago

@kidneyomicsamps @gratefull080504 @ColumbiaKidney Yes, this is a very real benefit. And it is moving fast over the past few years #NephJC

**Sarah Melville @sarahkmels**

4 days ago

RT @mcstarr1: @kidneyomicsamps @hswapnil @Maximal_Change @NephJC @rheault_m @CJASN Check it out here: <https://t.co/RmErTreKxg> #nephjc

**Michelle Starr @mcstarr1**

4 days ago

@kidneyomicsamps @gratefull080504 @Maximal_Change @ColumbiaKidney This is a really good point. The field has moved a long way in a short period of time!#nephjc

**Jennie Lin, MD MTR @jenniejlin**

4 days ago

@Maximal_Change @ccxjwang @NephJC it will get cheaper and cheaper when more competitors. i bet there will be more sequencing platform companies than dialysis companies 10 years from now #NephJC

**Nephrology Jrnl Club @NephJC**

4 days ago

T2I: Known or expected pathogenic variants were detected in the 59 ACMG medically actionable genes in 34 of the 2187 patients (1.6%) assessed. Although secondary these findings can have implications for nephrology care #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @NephJC: T2I: Known or expected pathogenic variants were detected in the 59 ACMG medically actionable genes in 34 of the 2187 patients (...)



Timothy Yau @Maximal_Change

4 days ago

@kidneyomicsamps @ColumbiaKidney @nainamdar @hswapnil @NephJC Paper quoted 1.4% in healthy population having pathologic variants after filtering #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@KidneyCathy original AURORA study consent did not offer option for return of results, but under new EU regulations, study participants can request their data, #nephjc



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T2D: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients....



Kevin J. Fowler @gratefull080504

4 days ago

@kidneyomicsamps : and the more that the #patientvoice supports innovation like NGS patient expectations must be elevated #nephJC



Nephrology Jrnl Club @NephJC

4 days ago

T2J: What to make of the results? What surprises you the most? #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@hswapnil @KidneyCathy agreed its tough ethically. ultimately i think combining research and CLIA-level sequencing will help reduce this issue #nephjc



David Ellison @dhekidney

4 days ago

@jenniejlin @Maximal_Change @ccxjwang @NephJC The problem IMHO is that we still need TREATMENTS if we find an Alport gene or a PKD gene. Other than avoiding an unnecessary treatment, we still aren't like cards where there are clearly beneficial treatments. #nephJC



Timothy Yau @Maximal_Change

4 days ago

@gratefull080504 We welcome their input! On the same team here, and we all want the best for the patient #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

#nephjc

John Montford, MD @JohnRMontford

4 days ago



@NephJC #nephjc given higher prevalence of PKD discovered wonder what GANAB mutation prevalence would be in similar cohort? Not among the candidate genes screened in the study. Always wonder about those: "looks like PKD but not really" patients

Krzysztof Kiryluk @kirylukk

4 days ago



@Maximal_Change @gratefull080504 and the prices continue to drop. I think the clinical interpretation is more expensive than the test itself at this point. Overall, cost probably getting comparable to an MRI scan... and I am confident that this will get much cheaper soon. #NephJC

Jennie Lin, MD MTR @jenniejlin

4 days ago



@NephJC "nephropathy of unknown origin" diagnosed finally for a subset of patients. #NephJC

Nephrology Jrnl Club @NephJC

4 days ago



@MarvinGonzlez16 @ssfouk @KidneyCathy Then do u use serological markers for routine diagnosis? #NephJC

Swapnil Hiremath, MD, MPH @hswapnil

4 days ago



RT @JohnRMontford: @NephJC #nephjc given higher prevalence of PKD discovered wonder what GANAB mutation prevalence would be in similar coho...

Matt Sampson @kidneyomicsamps

4 days ago



@kirylukk @Maximal_Change @gratefull080504 Totally. The real cost will be how to interpret all of this data #nephjc

Sarah Melville @sarahkmels

4 days ago



RT @gratefull080504: @Maximal_Change : Tim, I think people living with #kidneydisease + patient organizations need to weigh in on this issu...

Kevin J. Fowler @gratefull080504

4 days ago



We need radical change for the future of #nephrology as a profession + to transform #patientcare Long overdue #nephJC

**Katalin Susztak @KSusztak**

4 days ago

@Nephro_Sparks Did you change to APOL1? #NephJC

**Jennie Lin, MD MTR @jenniejlin**

4 days ago

@dhekidney @Maximal_Change @ccxjwang @NephJC Yeah, but we need more data to develop the treatments ... #NephJC

**Gary Singer @GarySingerMD**

4 days ago

@NephJC Also monogenetic forms of FSGS likely to be treatment resistant and less likely to recur post transplant #NephJC

**Timothy Yau @Maximal_Change**

4 days ago

@mcstarr1 @kidneyomicsamps @gratefull080504 @ColumbiaKidney Exactly. Six years ago, it was prohibitively difficult getting complement factor testing for aHUS, now it's a simple form with 4 week turnaround. #NephJC

**Aisha Shaikh @aishaikh**

4 days ago

Hi, this is Aisha Shaikh from Mount Sinai NYC. No COI #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

T3A: Limitations of exome sequencing listed: 1st-Suboptimal coverage of some clinically relevant regions, eg- the mitochondrial DNA or duplicated regions of PKD1. 2nd - Inability to detect intronic & copy-number variants. Does this underestimate the overall genetic burden? #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

@dhekidney @jenniejlin @Maximal_Change @ccxjwang @NephJC true. Still, as @rheault_m sez, good to give a name for unknown cause + Family/genetic counselling Avoidance of needless IS Transplant/donor counselling #NephJC

**Sarah Melville @sarahkmels**

4 days ago

RT @kidneyomicsamps: @gratefull080504 @Maximal_Change The more that papers are published like @ColumbiaKidney that show that testing matter...



Columbia Nephrology @ColumbiaKidney

4 days ago

@kidneyomicsamps @kirylukk @Maximal_Change @gratefull080504 well said. we can generate the data, but now we need to systematize how to best put it into practice... #nephjc



Matt Sparks @Nephro_Sparks

4 days ago

@kidneyomicsamps ((We)) as the scientific community need to place a ((large)) degree skepticism & rigor in assigning pathogenicity of rare variants as the implications could be large & devastating to people especially as the technology gets cheaper. #NephJC It's about more than a paper



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

great point #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

#NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@Nephro_Sparks @kidneyomicsamps That's why we take them to lab to validate #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

T3B: Any other limitations? Are these results generalizable to your population? #NephJC



Timothy Yau @Maximal_Change

4 days ago

@kidneyomicsamps @ColumbiaKidney @nainamdar @hswapnil @NephJC Just reading the abstract. 23% initially had a candidate pathologic mutation, but filtered down to 1.4% after filtering out previously reported variants #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@NephJC Yes! @ColumbiaKidney has also led publications showing a substantial burden of #CNV in children with any CKD and especially patients with CAKUT diagnoses, which are a major cause of CKD in kids #nephjc



Cynthia Wang @ccxjwang

4 days ago

@jenniejin @NephJC I agree, I think it will be great for those patients to have actually have a named diagnosis / etiology to their disease, and it's great that many of these diagnoses are actionable according to the results as well. #NephJC



Marvin Gonzalez @MarvinGonzlez16

4 days ago

@NephJC its incredible that 17% patients had #CKDu. #NephJC



Vighnesh Walavalkar @vighnesh_w

4 days ago

RT @NephJC: T0g: In adults, exome sequencing has been applied in patients with FSGS. Check out this amazing VA by @rheault_m @cjasn #NephJC...



Kevin J. Fowler @gratefull080504

4 days ago

@dhekidney : There are 12 companies are investing in #adpkd + several are investing in #alport Pharma is incentivized to find treatments for #raredisease #kidneydisease How much longer can patients accept a system that rewards renal failure? Enough #nephJC



Nephrology Jrnl Club @NephJC

4 days ago

T3C: Is there a risk of falsely attributing causality in few patients where genetic variants are not well specified? #NephJC



Jennie Lin, MD MTR @jenniejin

4 days ago

@NephJC I actually wonder if more patients in the community would be reclassified. #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @Nephro_Sparks: @kidneyomicsamps ((We)) as the scientific community need to place a ((large)) degree skepticism & rigor in assigning pat...



David Ellison @dhekidney

4 days ago

@Nephro_Sparks @kidneyomicsamps To really close the loop here, we still need effective treatments to tell us about pathogenesis. Sort of Koch's postulates for the 21st century. #NephJC

**Jennie Lin, MD MTR** @jenniejlin

4 days ago

@Nephro_Sparks @kidneyomicsamps I've been taking notes from the cardiologists. ;) #NephJC

**Matt Sparks** @Nephro_Sparks

4 days ago

RT @dhekidney: @Nephro_Sparks @kidneyomicsamps To really close the loop here, we still need effective treatments to tell us about pathogene...

**Kevin J. Fowler** @gratefull080504

4 days ago

I think they would #nephJC

**Columbia Nephrology** @ColumbiaKidney

4 days ago

@Nephro_Sparks @hswapnil @kidneyomicsamps you raise a great point. we use guidelines for interpretation but like any other medical test, genetic testing does not give absolute answers..certainty varies #nephjc

**David Ellison** @dhekidney

4 days ago

@gratefull080504 COI: I am on ASN Council, but this is what we spend a lot of time talking about. Hoping KHI and Kidney X help! #NephJC

**Matt Sampson** @kidneyomicsamps

4 days ago

@NephJC Yes, (1) there is this risk but also (2) given variable penetrance due to environ and/or genetic modifiers, we can't always be certain about what will happen in pts. This is less of a concern here b/c all already have CKD. But a warning for presymptomatic screening #nephjc

**Jennie Lin, MD MTR** @jenniejlin

4 days ago

@dhekidney @Nephro_Sparks @kidneyomicsamps I'm looking up Koch's postulates on Wikipedia. :) #NephJC

**Matt Sampson** @kidneyomicsamps

4 days ago

+100 #nephjc

**Columbia Nephrology** @ColumbiaKidney

4 days ago

@Nephro_Sparks @hswapnil @kidneyomicsamps would argue it's best to see things in a bayesian, probalistic framework #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

T3D: Which patients in your clinical practice do you think will benefit the most with exome sequencing? #NephJC

**Jung Hoon Son, M.D. @junghoon_sonMD**

4 days ago

@Nephro_Sparks Definitely. Early constitutional genomics often jumped at so-called pathogenic mutations leading to unintended pregnancies for some parents. Until select brave parents proceeded with pregnancies, having normal newborns. This effectively rewrote some gene pathogenicity #nephjc

**Krzysztof Kiryluk @kirylukk**

4 days ago

We are performing WES of all KPMP AKI cases recruited at Columbia... completely agree, need WES and preferably WGS for all KPMP participants... #NephJC

**Swapnil Hiremath, MD, MPH @hswapnil**

4 days ago

RT @kidneyomicsamps: @NephJC Yes, (1) there is this risk but also (2) given variable penetrance due to environ and/or genetic modifiers, we...

**Matt Sampson @kidneyomicsamps**

4 days ago

@ColumbiaKidney @Nephro_Sparks @hswapnil Agree. In other words "Genetics is not destiny. Continue to act like a doctor." #nephjc

**Katalin Susztak @KSusztak**

4 days ago

@jenniejlin @Nephro_Sparks @kidneyomicsamps Lab validation of coding or non-coding variants are absolutely essential, you remember we all have probably 200 loss of function variants on average, yet no disease, hard to know which is causing disease out of so many #NephJC

**Columbia Nephrology @ColumbiaKidney**

4 days ago

@Nephro_Sparks @hswapnil @kidneyomicsamps you incorporate the genetic testing results with the patient's clinical hx, findings, etc. and importantly reassess your conclusions over time to help support giving them the most accurate answer you can, at that time #nephjc

**Nephrology Jrnl Club @NephJC**

4 days ago

@ssfouk @KidneyCathy Sadly no such compulsion here to 'assign a diagnosis'. Hence it is not sought after often #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @kirylukk: We are performing WES of all KPMP AKI cases recruited at Columbia... completely agree, need WES and preferably WGS for all KP...



Mario Funes MD @MarioFunesMD

4 days ago

Do we know how many "healthy" patients without CKD have these genetic variants? #NephJC



Mira Krendel @KidneyMyosin

4 days ago

@PAWellingMD @Maximal_Change @kidneyomicsamps @ColumbiaKidney @nainamdar @hswapnil @NephJC Following this discussion with a lot of interest since we are currently working out ways to perform rapid functional testing in one of the #SRNS genes. #NephJC .



Columbia Nephrology @ColumbiaKidney

4 days ago

@KSusztak @jenniejlin @Nephro_Sparks @kidneyomicsamps that is a great point, i think developing high-throughput assays so that any new variant can be tested as part of the variant interpretation workflow will be key as genetic testing expands #nephjc



Matt Sparks @Nephro_Sparks

4 days ago

@ColumbiaKidney @hswapnil @kidneyomicsamps I'm more talking about in the future for other rare variants not described before... #NephJC



Rhona Bejarano @Rhona1885

4 days ago

Hi everyone, this is Rhona Bejarano venezuelan nephrologist, NSCM intern #NephJC



Gary Singer @GarySingerMD

4 days ago

@NephJC Late stage CKD of unknown etiology, steroid resistant FSGS #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: T2I: Known or expected pathogenic variants were detected in the 59 ACMG medically actionable genes in 34 of the 2187 patients (...)



Kevin J. Fowler @gratefull080504

4 days ago

I am not a nephrologist but in my opinion: #FSGS #igan #diabetickidneydisease #adpkd #alport #CKDu and anyone with a history of early kidney failure without the cause of #kidneyfailure #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@KSusztak @jenniejlin @Nephro_Sparks Agree and also have to take into account model system/assay...lack of concordance may not be indicative of true pathogenicity. Appeal for isogenic models #nephjc



Timothy Yau @Maximal_Change

4 days ago

@NephJC I think the atypical responders of glomerular disease, young patients who present with unknown advanced CKD are what immediately jump to my mind #NephJC



David Ellison @dhekidney

4 days ago

@kirylukk I guess I was wondering about whether finding a pathogenic variant should be an exclusion? #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @ColumbiaKidney: @Nephro_Sparks @hswapnil @kidneyomicsamps you incorporate the genetic testing results with the patient's clinical hx, f...



Nephrology Jrnl Club @NephJC

4 days ago

What are the benefits of knowing the genetic diagnosis in your patients? In my population the benefits still dont justify the cost of testing in all patients. Hoping it to come down #NephJC



Cathy Quinlan @KidneyCathy

4 days ago

RT @KSusztak: @jenniejlin @Nephro_Sparks @kidneyomicsamps Lab validation of coding or non-coding variants are absolutely essential, you rem...



Matt Sparks @Nephro_Sparks

4 days ago

RT @KSusztak: @jenniejlin @Nephro_Sparks @kidneyomicsamps Lab validation of coding or non-coding variants are absolutely essential, you rem...



Jennie Lin, MD MTR @jenniejlin

4 days ago

@kidneyomicsamps @KSusztak @Nephro_Sparks Then there's the question of readout for kidney. For dilated cardiomyopathy genetics, patch-clamping iPSC-cardiomyocytes gives disease-relevant readout. #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

#nephjc



Sarah Melville @sarahkmels

4 days ago

@NephJC seems like a low yield, yes? #nephjc



Timothy Yau @Maximal_Change

4 days ago

@KSusztak @jenniejlin @Nephro_Sparks @kidneyomicsamps 200? But eventually @Nephro_Sparks and @jenniejlin can just CRISPR those back in for me, right? #NephJC



Gary Singer @GarySingerMD

4 days ago

@NephJC Any role in assessment of potential kidney donors? #NephJC



Cynthia Wang @ccxjwang

4 days ago

@NephJC I think there is a small risk. Some patients could have these pathogenic variants but their CKD did not necessarily result from that genetic variant. In fact, could there be cases where they have these less studied variants but these were incidental to their own CKD? #NephJC



Sarah Melville @sarahkmels

4 days ago

@mcstarr1 @kidneyomicsamps @gratefull080504 @Maximal_Change @ColumbiaKidney oops, #nephjc ^



Paul A Welling @PAWellingMD

4 days ago

@dhekidney @NephJC #NephJC surprised it isn't' apart of the KPMP workup



Matt Sampson @kidneyomicsamps

4 days ago

@jenniejlin @KSusztak @Nephro_Sparks Totally agree. Appropriate read-out is key #nephjc



Columbia Nephrology @ColumbiaKidney

4 days ago

@NephJC great question, i think it comes down to having more a specific idea of the cause of disease you can better direct patients and families to the appropriate treatment in many cases. #nephjc



Matt Sparks @Nephro_Sparks

4 days ago

@Maximal_Change @KSusztak @jenniejlin @kidneyomicsamps But maybe another mutation arises even worse #NephJC



Marvin Gonzalez @MarvinGonzlez16

4 days ago

@MarioFunesMD Good point @MarioFunesMD #NephJC #NSMC2019



Fitsum Tilahun MD @fitse_t

4 days ago

RT @NephJC: T2G: 62% of patients with COL4A3, COL4A4, or COL4A5 mutations did not have clinical diagnoses of Alport syndrome or thin baseme...



Luis Enrique Alvarez @LuisEnr80631682

4 days ago

Hello everyone!! Here Luis Alvarez. Nephrologist from KidneyTeamLaRaza. Mexico City. #NephJC No COI



Fitsum Tilahun MD @fitse_t

4 days ago

RT @NephJC: T2H: In 15 patients (16%) with a clinical diagnosis of focal segmental glomerulosclerosis, the genetic diagnosis could disfavor...



Krzysztof Kiryluk @kirylukk

4 days ago

@KSusztak Agree, some genes are tolerant of LOF, but there rare many LOF variants are known to be pathogenic and are diagnostic with no need for further validation. Novel variants in novel genes is a whole different game... #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

Amen Isn't that the role of medicine + science to improve the suffering of people living with #kidneydisease? Kudos again to @ColumbiaKidney for their work I really appreciate it #NephJC



Katalin Susztak @KSusztak

4 days ago

@Maximal_Change @jenniejlin @Nephro_Sparks @kidneyomicsamps I am still a BIG believer of model organisms... mice (CRISPR mice) #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@NephJC and even if treatment is not yet available, w/no clinical trials yet, recognizing patients who have a specific disease subtype is the first step to developing those targeted treatments #nephjc



Mario Funes MD @MarioFunesMD

4 days ago

How about a risk assessment tool that would guide which patients would benefit the most from genetic testing? #NephJC



Marvin Gonzalez @MarvinGonzlez16

4 days ago

@neforantes @opsoms @ISNeducation @ReviewMedicc Hi Carlos @neforantes #NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@Nephro_Sparks @Maximal_Change @KSusztak @kidneyomicsamps That's a whole separate #NephJC chat for the future



Nephrology Jrnl Club @NephJC

4 days ago

T3E: Last few minutes left. What are your take home points? Final thoughts/comments? #NephJC



Gates Colbert, MD @DoctorGates

4 days ago

@NephJC We need to not just live in the present. But realize our patients have decades of life ahead. Let's start with as specific a diagnosis as possible, so we can apply future treatments to come. #nephjc



Timothy Yau @Maximal_Change

4 days ago

@NephJC I had a patient with an Alport's-like biopsy and strong family history where it became incredibly important when it came to her current pregnancy, risk for her child, etc. Was invaluable for her care #NephJC



Matt Sampson @kidneyomicsamps

4 days ago

@ColumbiaKidney @NephJC Should also allow for genomically stratified clinical trials #nephjc



Columbia Nephrology @ColumbiaKidney

4 days ago

@MarioFunesMD yes! we need all minds and fields on deck to help inform use. i see a lot of opportunities for incorporation of exome ehr, and other data to help guide the creation of such a tool #nephjc



Cynthia Wang @ccxjwang

4 days ago

@NephJC Perhaps an option is to do 1) targeted gene sequencing of the most common nephropathy-related genes (cheaper and more efficient) and 2) specifically in certain subsets that could benefit the most. Such as those with family hx of CKD or those with unknown diagnoses. #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

@ColumbiaKidney : I said this earlier but no one seemed to like my suggestion #NephJC



Sarah Melville @sarahkmels

4 days ago

@LBEBEN hmm, yes, perhaps.. #nephjc ughh I keep forgetting the # tonight-distracted... :(



Nephrology Jrnl Club @NephJC

4 days ago

RT @ColumbiaKidney: @NephJC and even if treatment is not yet available, w/no clinical trials yet, recognizing patients who have a specific...



John Montford, MD @JohnRMontford

4 days ago

@NephJC The folks I round on in the HD unit that I code "I dunno" on the cms-2728 form #nephjc



Katalin Susztak @KSusztak

4 days ago

@kirylukk Yes, but most of these variants in our WES biobanks are ultrarare (so novel) no? #NephJC



Jennie Lin, MD MTR @jenniejlin

4 days ago

@KSusztak @Maximal_Change @Nephro_Sparks @kidneyomicsamps Just as sequencing costs go down, I wish mouse costs would go down too! Any humanized kidney mouse models? #NephJC



Vanessa Lerma @VanessaLerma17

4 days ago

@NephJC Patients with Nephropathy of unknown origin not candidates for biopsy #nephJC



Matt Sparks @Nephro_Sparks

4 days ago

@KSusztak @Maximal_Change @jenniejlin @kidneyomicsamps Taking to mammalian organism is expensive and potential pitfalls. But agree it is important step. When TRPC6 found it was a real potential target as gain of function and ion channel #NephJC



Mario Funes MD @MarioFunesMD

4 days ago

@ColumbiaKidney Yes, I agree. #NephJC



Katalin Susztak @KSusztak

4 days ago

@jenniejlin @Maximal_Change @Nephro_Sparks @kidneyomicsamps I agree right now the hardest is not sequencing and informatics, but validation \$\$\$+time #NephJC



Sarah Melville @sarahkmels

4 days ago

@Nephro_Sparks #nephjc ^ ughh I keep forgetting this # tonight... distracted or something off my game - rookie mistake :(



Marvin Gonzalez @MarvinGonzlez16

4 days ago

What about in patient with known low birth weight? #NephJC #NSMC2019



Arshad Ali @docarshadali

4 days ago

RT @ColumbiaKidney: @NephJC and even if treatment is not yet available, w/no clinical trials yet, recognizing patients who have a specific...



Jennie Lin, MD MTR @jenniejlin

4 days ago

@gratefull080504 @ColumbiaKidney Kevin, I think your tweet just got lost in the shuffle of the sheer volume of tweets happening tonight! #NephJC

Timothy Yau @Maximal_Change

4 days ago



@ccxjwang @NephJC Yes, but if we build up the database with enough of these variants, machine learning will (hopefully) be able to tell us which are pathologic, and which are incidental. Basically I think we need to do this on more people and have strength in numbers #NephJC

David Ellison @dhekidney

4 days ago



@KSusztak @Maximal_Change @jenniejlin @Nephro_Sparks @kidneyomicsamps and unlike some other diseases and organs, a number of mouse models of kidney disease DO provide strong mechanistic insight IMHO #NephJC

Sarah Melville @sarahkmels

4 days ago



RT @kidneyomicsamps: @NephJC Yes! @ColumbiaKidney has also led publications showing a substantial burden of #CNV in children with any CKD a...

Neeraj Inamdar @nainamdar

4 days ago



@NephJC @MarioFunesMD Ya that was nice abstract.. Potential candidate genes associated with CKDu thr GWAS.. Significantly high risk association with NLRP4 and PRKN genes.. #NephJC

Katalin Susztak @KSusztak

4 days ago



@Nephro_Sparks @Maximal_Change @jenniejlin @kidneyomicsamps Another great example #NephJC

Matt Sparks @Nephro_Sparks

4 days ago



@KSusztak @Maximal_Change @jenniejlin @kidneyomicsamps Still no therapy and look at what TRPC6 KO in Akita Diabetes does (worsens) #NephJC
<https://t.co/ZoVuLItA3P>

Sarah Melville @sarahkmels

4 days ago



RT @gratefull080504: @dhekidney : There are 12 companies are investing in #adpkd + several are investing in #alport Pharma is incentivized...

Jung Hoon Son, M.D. @junghoon_sonMD

4 days ago



@Maximal_Change @ccxjwang @NephJC Wish we could aggregate all the CKD genomics into one place... #nephjc imagine!



Matt Sampson @kidneyomicsamps

4 days ago

@Maximal_Change @ccxjwang @NephJC On a research basis, I think that every kidney patient should undergo WES and SNP genotyping to achieve what you are suggesting. And I think it would work #nephjc



Nephrology Jrnl Club @NephJC

4 days ago

@yasar_caliskan Would you screen all donors or those only with a known genetic disease in the proband? #NephJC



David Ellison @dhekidney

4 days ago

@VanessaLerma17 @NephJC This is the goal of KPMP! #NephJC



Susan Thomas @kidsbeansdoc

4 days ago

These patients will go onto transplantation. Avoiding unnecessary immunosuppression (and complications) will improve QoL for many.#nephjc



Columbia Nephrology @ColumbiaKidney

4 days ago

@KSusztak @jenniejlin @Maximal_Change @Nephro_Sparks @kidneyomicsamps yes, agree. i hope ours and other sequencing studies help this effort insofar that once genetic sequencing is more widely used, there's greater incentive to make it more sophisticated by developing high-throughput f/u assays to add into the pipeline #nephjc



Cathy Quinlan @KidneyCathy

4 days ago

RT @gratefull080504: @ColumbiaKidney : I said this earlier but no one seemed to like my suggestion #NephJC <https://t.co/nVgQrqt5>



Tiffany Caza @Tiff_Caza

4 days ago

@NephJC If this becomes prime time and we start doing this testing, how should we best share our data to learn more about individual variants? What databases are recommended? #NephJC



Timothy Yau @Maximal_Change

4 days ago

@ColumbiaKidney @NephJC I feel like this is probably where heme-onc was 25 years ago when they started doing cytogenetics on their bone marrow. Now they have targeted therapy, this is hopefully the nephrology equivalent #NephJC

**Sarah Melville** @sarahkmels

4 days ago

@NephJC yes, thanks- like a false positive? #nephjc

**Sarah Melville** @sarahkmels

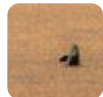
4 days ago

RT @kidneyomicsamps: @NephJC Yes, (1) there is this risk but also (2) given variable penetrance due to environ and/or genetic modifiers, we...

**Swapnil Hiremath, MD, MPH** @hswapnil

4 days ago

RT @Maximal_Change: @ColumbiaKidney @NephJC I feel like this is probably where heme-onc was 25 years ago when they started doing cytogeneti...

**Susan Thomas** @kidsbeansdoc

4 days ago

RT @kidneyomicsamps: @Maximal_Change @ccxjwang @NephJC On a research basis, I think that every kidney patient should undergo WES and SNP ge...

**Kevin J. Fowler** @gratefull080504

4 days ago

#nephjc We are willing to spend over \$34 billion for #dialysis that does not fully rehabilitate a person living with #kidneydisease but some people are questioning the cost of NGS? And people are surprised with my frustration? #NephJC

**Paul A Welling** @PAWellingMD

4 days ago

RT @kidneyomicsamps: @Maximal_Change @ccxjwang @NephJC On a research basis, I think that every kidney patient should undergo WES and SNP ge...

**Jennie Lin, MD MTR** @jenniejlin

4 days ago

@dhekidney @KSusztak @Maximal_Change @Nephro_Sparks @kidneyomicsamps dish can't replace physiology, but it would be great to have mice with full-grown (not iPSC-injected) kidneys with human genome to interrogate #NephJC

**Nephrology Jrnl Club** @NephJC

4 days ago

#NephForward #NephJC

**Neeraj Inamdar** @nainamdar

4 days ago

RT @ColumbiaKidney: @NephJC and even if treatment is not yet available, w/no clinical trials yet, recognizing patients who have a specific...



Jung Hoon Son, M.D. @junghoon_sonMD

4 days ago

RT @KSusztak: @jenniejlin @Nephro_Sparks @kidneyomicsamps Lab validation of coding or non-coding variants are absolutely essential, you rem...



Sarah Melville @sarahkmels

4 days ago

RT @Nephro_Sparks: @kidneyomicsamps ((We)) as the scientific community need to place a ((large)) degree skepticism & rigor in assigning pat...



Cathy Quinlan @KidneyCathy

4 days ago

RT @ccxjwang: @NephJC I think there is a small risk. Some patients could have these pathogenic variants but their CKD did not necessarily r...



Matt Sampson @kidneyomicsamps

4 days ago

@Tiff_Caza @NephJC Such an important comment & idea. We need to bring this data out to the community to make collective discoveries, while also respecting patient privacy. @KSusztak & I have both made online databases to share genetic and expression information. More of this has to happen #nephjc



Matt Sampson @kidneyomicsamps

4 days ago

RT @jenniejlin: @dhekidney @KSusztak @Maximal_Change @Nephro_Sparks @kidneyomicsamps dish can't replace physiology, but it would be great t...



Kevin J. Fowler @gratefull080504

4 days ago

@Maximal_Change Dead on Tim I wish everyone would read the book "Emperor of All Maladies" to learn how oncology became the field it is today #nephrology can do the same #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @kidneyomicsamps: @Tiff_Caza @NephJC Such an important comment & idea. We need to bring this data out to the community to make collectiv...



Gary Singer @GarySingerMD

4 days ago

@gratefull080504 Agree - need to educate insurers that spending \$ on NGS now will save \$ later and improve patients' lives #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@kushjaya @jenniejlin @NephJC we manually curated OMIM, <https://t.co/lpIBWshHFm>, two large gene-disease databases for genes associated w/mendelian forms of CKD #nephjc



Susan Thomas @kidsbeansdoc

4 days ago

RT @kidneyomicsamps: @Tiff_Caza @NephJC Such an important comment & idea. We need to bring this data out to the community to make collectiv...



Matt Sampson @kidneyomicsamps

4 days ago

RT @GarySingerMD: @gratefull080504 Agree - need to educate insurers that spending \$ on NGS now will save \$ later and improve patients' live...



Fitsum Tilahun MD @fitse_t

4 days ago

RT @Maximal_Change: @ColumbiaKidney @NephJC I feel like this is probably where heme-onc was 25 years ago when they started doing cytogeneti...



Krzysztof Kiryluk @kirylukk

4 days ago

@dhekidney Would be hard to sequence everyone with AKI before a biopsy, but certainly finding a pathogenic variant provides a valuable piece of information that could lead to exclusion from some KPMP projects, but also maybe inclusion in others? #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

Thanks to @ @yasar_caliskan and @ @jenniejlin for the amazing summary. And @rheault_m for the commentary. This made understanding it so easy for novice like me #NephJC



Sarah Melville @sarahkmels

4 days ago

RT @ColumbiaKidney: @Nephro_Sparks @hswapnil @kidneyomicsamps would argue it's best to see things in a bayesian, probalistic framework #nep...



Columbia Nephrology @ColumbiaKidney

4 days ago

@kushjaya @jenniejlin @NephJC this is a standard approach for most clinical diagnostic sequencing - you look at genes reported to be associated with your disease of interest/clinical indication for testing #nephjc



Suneel Udani @CaptainKidney79

4 days ago

RT @Maximal_Change: @ColumbiaKidney @NephJC I feel like this is probably where heme-onc was 25 years ago when they started doing cytogeneti...



Sarah Melville @sarahkmels

4 days ago

RT @kidneyomicsamps: @ColumbiaKidney @Nephro_Sparks @hswapnil Agree. In other words "Genetics is not destiny. Continue to act like a doctor..."



David Ellison @dhekidney

4 days ago

@NephJC Yes, but I am much more hopeful about single gene pathologies than just 'precision medicine' like much onc. See the recent JCI review by Joyner. We need solid science and not hype. #NephJC



Columbia Nephrology @ColumbiaKidney

4 days ago

@kushjaya @jenniejlin @NephJC see Methods in the Appendix 1 for a summary, and our annals paper for a longer, more in-depth discussion: <https://t.co/G4QCRrWWk1> #nephjc



Timothy Yau @Maximal_Change

4 days ago

@gratefull080504 You're the second person to mention this book to me this week, I'll have to look into it #NephJC



Matt Sparks @Nephro_Sparks

4 days ago

Fantastic study @ColumbiaKidney and thank you for joining. This is an important study and shows the importance of genetic basis of kidney disease. I hope @NIH was watching... time to 💰💰💰 the funding. The time is now... thanks @jenniejlin @divyaa24 @yasar_caliskan #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

If you haven't, please sign up for our weekly newsletter <https://t.co/5aumn8qL85> #NephJC



Gary Singer @GarySingerMD

4 days ago

@NephJC @yasar_caliskan @jenniejlin @rheault_m Great summary and discussion #NephForward #NephJC



Kevin J. Fowler @gratefull080504

4 days ago

@dhekidney : I have a lot of confidence of senior leadership of @ASN + @Kidney_X are really trying to transform the field I respect them as well #nephJC



Matt Sampson @kidneyomicsamps

4 days ago

RT @Nephro_Sparks: Fantastic study @ColumbiaKidney and thank you for joining. This is an important study and shows the importance of geneti...



Paul A Welling @PAWellingMD

4 days ago

@jenniejlin @dhekidney @KSusztak @Maximal_Change @Nephro_Sparks @kidneyomicsamps #NephJC I like @KSusztak 's idea of just going all Crispr on mouse, realizing that it will take at least 12-18 months. As these are all rare, need to think about common pathogenic (and drugable) pathways.



Gary Singer @GarySingerMD

4 days ago

RT @Nephro_Sparks: Fantastic study @ColumbiaKidney and thank you for joining. This is an important study and shows the importance of geneti...



Sarah Melville @sarahkmels

4 days ago

@kidneyomicsamps @ColumbiaKidney @Nephro_Sparks @hswapnil yes, agreed, "just good old fashioned medicine" (& without hype) #nephjc cc: Dr. @thebyrdlab



Columbia Nephrology @ColumbiaKidney

4 days ago

@Nephro_Sparks @NIH @jenniejlin @divyaa24 @yasar_caliskan thank you! #nephjc



Timothy Yau @Maximal_Change

4 days ago

@NephJC @yasar_caliskan @jenniejlin @rheault_m Thanks for curating! Great work on the visual abstract, summaries, commentary, etc. Great chat #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

Thanks for joining us tonight and for contributing to the discussion. There are 2 more exciting chats coming up on the same topic on wednesday at 9pm IST and 8pm GMT #NephJC



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: If you haven't, please sign up for our weekly newsletter
<https://t.co/5aumn8qL85> #NephJC



Nephrology Jrnl Club @NephJC

4 days ago

Would you like to have some #NephJC swag? Order it here <https://t.co/eTUrZY27Qo>



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

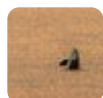
Thanks to authors @ColumbiaKidney & @kirylyuk + @kidneyomicsamps @junghoon @ccxjwang and @Maximal_Change and all of you for bringing their A game to tonight's #nephjc



Matt Sampson @kidneyomicsamps

4 days ago

@PAWellingMD @jenniejlin @dhekidney @KSusztak @Maximal_Change @Nephro_Sparks I like this idea as well. And regulatory regions, genetic background and environmental exposures also matter in modulating penetrance. Lots of complementary work to do #nephjc



Susan Thomas @kidsbeansdoc

4 days ago

Great turn out and discussion. #nephjc



Timothy Yau @Maximal_Change

4 days ago

@ColumbiaKidney @Nephro_Sparks @NIH @jenniejlin @divyaa24 @yasar_caliskan
Thanks for chatting, always great having the author(s) join #NephJC



Cathy Quinlan @KidneyCathy

4 days ago

Thank you for joining us Emily - it's been fascinating, I look forward to seeing more from your group @ColumbiaKidney #nephjc



Kevin J. Fowler @gratefull080504


4 days ago

#nephjc Have to run Great paper tonight to discuss + has me very motivated Thank you @ColumbiaKidney for your important research



Matt Sparks @Nephro_Sparks

4 days ago

The genetics numbers: 535 Tweets. 102 Participants. February 11th 2019. More #NephJC  here <https://t.co/J96TdwAiSB> via @symplur


Nephrology Jrnl Club @NephJC

4 days ago

Time to sign off! Please feel free to continue sharing your thoughts with #NephJC Good bye and Good night


Columbia Nephrology @ColumbiaKidney

4 days ago

@gratefull080504 thank you, Kevin! this was awesome! #nephjc


Sarah Melville @sarahkmels

4 days ago

RT @NephJC: What are the benefits of knowing the genetic diagnosis in your patients? In my population the benefits still dont justify the c...


Sri Lekha Tummalapalli, MD, MBA @LTummalapalli

4 days ago

Consider innovative ways to finance sequencing beyond conventional medical reimbursement - e.g. social impact bonds. Insurance system will be slow to respond and may not reap the incentives. #nephjc


Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: Would you like to have some #NephJC swag? Order it here <https://t.co/eTUrZY27Qo>


Katalin Susztak @KSusztak

4 days ago

@PAWellingMD @jenniejlin @dhekidney @Maximal_Change @Nephro_Sparks @kidneyomicsamps Good point. We do not have many common pathogenic mutations. If we want precision medicine APOL1 or PKD1 can serve as an example. Common and highly penetrant coding variants. For rare variants it will be much much harder unless we develop better high-throughput methods #NephJC


Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

RT @NephJC: Thanks for joining us tonight and for contributing to the discussion. There are 2 more exciting chats coming up on the same top...


Matt Sampson @kidneyomicsamps

4 days ago

RT @KSusztak: @PAWellingMD @jenniejlin @dhekidney @Maximal_Change @Nephro_Sparks @kidneyomicsamps Good point. We do not have many common pa...

**Nephrology Jrnl Club @NephJC**

4 days ago

Special thanks to all the authors who joined the chat #NephJC

**Jung Hoon Son, M.D. @junghoon_sonMD**

4 days ago

RT @hswapnil: Thanks to authors @ColumbiaKidney & @kirylukk + @kidneyomicsamps @junghoon @ccxjwang and @Maximal_Change and all of you for b...

**Brittany J. Navarre @BritMasterFlash**

4 days ago

RT @Maximal_Change: @NephJC I think the atypical responders of glomerular disease, young patients who present with unknown advanced CKD are...

**Neeraj Inamdar @nainamdar**

4 days ago

@hswapnil @ColumbiaKidney @kirylukk @kidneyomicsamps @junghoon @ccxjwang @Maximal_Change Thanks a lot.. Primary author into discussion makes huge difference and a lot to gain from.. #nephJC

**Kevin J. Fowler @gratefull080504**

4 days ago

@ColumbiaKidney Thank you for joining + your important research #nephJC

**Sarah Melville @sarahkmels**

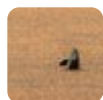
4 days ago

RT @NephJC: @yasar_caliskan Would you screen all donors or those only with a known genetic disease in the proband? #NephJC

**Sarah Melville @sarahkmels**

4 days ago

RT @Nephro_Sparks: @KSusztak @Maximal_Change @jenniejlin @kidneyomicsamps Still no therapy and look at what TRPC6 KO in Akita Diabetes does...

**Susan Thomas @kidsbeansdoc**

4 days ago

RT @kidneyomicsamps: @PAWellingMD @jenniejlin @dhekidney @KSusztak @Maximal_Change @Nephro_Sparks I like this idea as well. And regulatory...

**Sarah Melville @sarahkmels**

4 days ago

RT @gratefull080504: #nephjc We are willing to spend over \$34 billion for #dialysis that does not fully rehabilitate a person living with...



Susan Thomas @kidsbeansdoc

4 days ago

RT @Nephro_Sparks: Fantastic study @ColumbiaKidney and thank you for joining. This is an important study and shows the importance of geneti...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

Sorry @junghoon_sonMD ! Fastfingers #nephjc



Sarah Melville @sarahkmels

4 days ago

RT @PAWellingMD: @jenniejlin @dhekidney @KSusztak @Maximal_Change @Nephro_Sparks @kidneyomicsamps #NephJC I like @KSusztak 's idea of just...



Sarah Melville @sarahkmels

4 days ago

RT @NephJC: Would you like to have some #NephJC swag? Order it here <https://t.co/eTUrZY27Qo>



Michelle Rheault @rheault_m

4 days ago

@NephJC @CJASN Just arrived home from the @CureGn meeting in DC. What did I miss? #NephJC



Sarah Melville @sarahkmels

4 days ago

@NephJC Thank you for a great discussion & all of the refs. too on this important topic... #nephjc - TFW, so much great info. to absorb just signing off now, but chat ended 10 mins ago, lol, ;)



Sarah Melville @sarahkmels

4 days ago

@NephJC Thank you @ColumbiaKidney for the great work, ;) #nephjc



Sarah Melville @sarahkmels

4 days ago





RT @LTummalapalli: Consider innovative ways to finance sequencing beyond conventional medical reimbursement - e.g. social impact bonds. In...



Swapnil Hiremath, MD, MPH @hswapnil

4 days ago

@rheault_m @NephJC @CJASN @CureGn 109 people tweeting at once #NephJc <https://t.co/CK0uzJ7qdK>

-
-  **Itunu @itunuowoyemi** 4 days ago
Nicely summarized commentary on tonight's #NephJC by @rheault_m @NephJC Genetic Testing for Chronic Kidney Disease? Yes, please <https://t.co/qvMr7hxtiH>
-
-  **parvaneh abtahi @abtahi_parvaneh** 4 days ago
@NephJC It will be difficult to convince pts to get genetic testing without being able to offer a treatment. #nephjc
-
-  **Jung Hoon Son, M.D. @junghoon_sonMD** 4 days ago
Pretty cool having two coauthors representing for #nephjc @ColumbiaKidney @kirylukk! 🙌 to @NephJC
-
-  **Matt Sampson @kidneyomicsamps** 4 days ago
@sarahkmels @ColumbiaKidney @Nephro_Sparks @hswapnil @thebyrdlab I would amend that to "modern medicine based on old-fashioned principles" #NephJC

Showing 1 to 536 of 536 entries

 #NephJC content from Twitter.

Free Custom Analytics and Transcripts for #NephJC

100,000 Tweets

Symplur has over 100,000 #NephJC tweets going back to April 2014.
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