

#NephJC Transcript

Healthcare social media transcript of the #NephJC hashtag.

Wed, February 13th 2019, 2:00PM – Wed, February 13th 2019, 5:00PM (America/Toronto).

See #NephJC Influencers/Analytics.



Alex Hamilton @_ajhamilton

3 days ago

Trying to be ready for #NephJC at 2000... <https://t.co/d7gqjX82ZK>



Cathy Quinlan @KidneyCathy

3 days ago

Really looking forward to the European viewpoint on #NephJC And hoping there'll be authors online this time too? @ColumbiaKidney



Rob Peel @rabpeel

3 days ago

I'm ready for it! #nephjc



Tom Oates @toates_19

3 days ago

Or maybe fewer now #NephJC <https://t.co/16rjpfWR4z>



Matt Bottomley @bottomleymatt

3 days ago

Matt Bottomley. ACL/Renal SpR Oxford. Confess to know v little about genetics. No COI. #nephjc



Dr MK #NephrologistOnICU @_Dr_MK

3 days ago


@NephJC counting down the minutes to #nephjc!! Hope @johnasayer @gary_leggatt @RLWczyk @RenalRadar @RenalUCL @ICH_Nephro @KateBramham @kidgenaustralia @Shalabh_Sriv might join!





Nephrology Jrnl Club @NephJC


3 days ago

Good evening everyone! It's 08:00PM which means it's time for #NephJC. Welcome to tonight's discussion.


 **Nephrology Jrnl Club @NephJC** 3 days ago
I'm Laura Slattery (@Slatts_1) and I'll be your host tonight as we discuss this important topic. I'm an adult Nephrology trainee from Ireland, #NSMC faculty #NephJC.


 **Nephrology Jrnl Club @NephJC** 3 days ago
Please introduce yourself (even if you're just lurking and following along) and declare any Conflicts of Interests. Don't forget to use the hashtag! #NephJC.


 **Rob Peel @rabpeel** 3 days ago
Rab a bizzarley mild Highland Scotland, no COI #nephjc


 **Tom Oates @toates_19** 3 days ago
Tom, nephrologist from London #NephJC No COI Proud to work @NHSBartsHealth. Alas not at #bhheroes2019 <https://t.co/0I0Z2Q7Cxi>


 **Nephrology Jrnl Club @NephJC** 3 days ago
@rabpeel Welcome! #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
The paper we will be discussing tonight is "Diagnostic Utility of Exome Sequencing for Kidney Disease" published in NEJM in January of this year. #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
From an adult nephrology perspective, I'm particularly interested in exploring whether these technologies can help give an answer for the dreaded "CKD of unknown aetiology". #NephJC.

 **Nephrology Jrnl Club @NephJC** 3 days ago
@bottomleymatt Welcome Matt. You're not alone there! #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
@toates_19 @NHSBartsHealth Welcome Tom #NephJC

 **Eric Au @ericau** 3 days ago
@NephJC #nephjc hi Eric - nephrologist from Sydney. #nsmc intern. No COI.

Nephrology Jrnl Club @NephJC

3 days ago



Abbreviations that will be used throughout #NephJC tonight: •NGS: next generation sequencing •WES: whole exome sequencing •WGS: whole genome sequencing

Sinead Stoneman @Stones__

3 days ago



Hi all, Sinead, Irish Nephrology trainee, also part of #nsmc. No COI #NephJC

Alex Hamilton @_ajhamilton

3 days ago



Alex Hamilton, CL in Bristol. COI: my wife did an MD in HNF1B and in an earlier life I described an HNF4A mutation causing Fanconi syndrome. We love genetics 😊
#NephJC

Dr MK #NephrologistOnICU @_Dr_MK

3 days ago



@NephJC Hello #NephJC! Nephrologist basking in the glorious dawn of #genetic #nephrology <https://t.co/yKcpM81VSH>

Nephrology Jrnl Club @NephJC

3 days ago



A particular welcome to the new #NSMC interns joining in tonight (@NSMCInternship)
#NephJC

Nephrology Jrnl Club @NephJC

3 days ago



For anyone new or in need of a quick refresher, here is the quick guide on “How to #NephJC” <https://t.co/sxGJ0PD9zn>

Nephrology Jrnl Club @NephJC

3 days ago



@ericau Welcome Eric! Good to have you on board #NephJC

Mark Jesky @MarkJesky

3 days ago



Evening, Mark, nephrologist, Nottingham, no COI #nephjc

Alex Hamilton @_ajhamilton

3 days ago



RT @NephJC: A particular welcome to the new #NSMC interns joining in tonight (@NSMCInternship) #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@_Dr_MK @CityCorpHeath Welcome! Envious of the view #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

Don't have time to comb through the entire paper and supplementary material? Or just want the salient points? Check out the excellent summary by @yasar_caliskan NSMCIntern and @jenniejlin <https://t.co/HRfctGfo1b> and commentary by @rheault_m <https://t.co/RZO743DxFw> #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@MarkJesky Welcome Mark #NephJC



Cathy Quinlan @KidneyCathy

3 days ago

Cathy Quinlan. Paed nephjc melbourne. COI involves in a similar study. Lurking as getting ready for work. #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

Here is the brilliant visual abstract by Divya Bajpai (@divyaa24), who has already hosted the earlier thought provoking NephJCs on this paper #NephJC <https://t.co/ygs4I1VhJh>



Charlie Tomson @CharlieTomson

3 days ago

#NephJC Evening everyone. No conflicts of interest.



Nephrology Jrnl Club @NephJC

3 days ago

@KidneyCathy Welcome Cathy, great to have the expertise for the discussion. Lurking is always good #NephJC



Dr MK #NephrologistOnICU @_Dr_MK

3 days ago

@KidneyCathy An #exome from the morning ;) #NephJC



Cathy Quinlan @KidneyCathy

3 days ago

RT @_ajhamilton: Alex Hamilton, CL in Bristol. COI: my wife did an MD in HNF1B and in an earlier life I described an HNF4A mutation causin...



Nephrology Jrnl Club @NephJC

3 days ago

Without further ado...onto the background #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T0: There is ever increasing focus on genetic testing as a both a research and a clinical diagnostic modality throughout the medical field. #NephJC



Dr Ruth Silverton @DRuthclaire

3 days ago

#NephJC Ruth, nephrology specialist doc and teaching fellow @RoyalFreeNHS vague recollection of DNA. No COI



Cathy Quinlan @KidneyCathy

3 days ago

RT @NephJC: For anyone new or in need of a quick refresher, here is the quick guide on "How to #NephJC" <https://t.co/sxGJ0PD9zn>



Nephrology Jrnl Club @NephJC

3 days ago

@_ajhamilton Welcome Alex! Excellent :) #NephJC



Dr MK #NephrologistOnICU @_Dr_MK

3 days ago

@KidneyCathy HELLOOOO CATHY! *waves frantically* #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@Stones__ Welcome Sinead! #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T0: Genetic testing is complex as there is rarely once single mutation leading to the disease phenotype. DNA sequence variation can occur throughout the genome including insertions, deletions and single-nucleotide polymorphisms. #NephJC



Charlie Tomson @CharlieTomson

3 days ago

@NephJC @divyaa24 #nephjc it all depends on what we mean by 'medically actionable'! Great graphic.

**Hugo Diniz** @HDiniz_

3 days ago

Hello everyone! Nephrology fellow from Hospital de São João, Oporto, Portugal! No COI. Genetics-naive. This should be a good one. #NSMC intern #NephJC

**Clara Day** @ClaraDay13

3 days ago

Clara Nephrologist Birmingham #NephJC COI drowning not waving in ocean of admin so multitasking

**Tom Oates** @toates_19

3 days ago

@CharlieTomson @NephJC @divyaa24 The key point 8 minutes in #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

@DRuthclaire @RoyalFreeNHS Welcome Ruth! #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

T0: NGS has the ability to read many nucleotide bases at the same time, and can be applied to RNA sequencing, WES and WGS. The ability to flag uncatalogued DNA variants, high throughput and relatively modest cause make them attractive for use in clinical settings #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

@HDiniz_ Welcome Hugo! #NephJC

**Dr MK #NephrologistOnICU** @_Dr_MK

3 days ago

@rabpeel @NephJC @CityCorpHeath I see your tree, and I hold at - dead trunk #nephjc <https://t.co/r6WGELwznZ>

**Hugo Diniz** @HDiniz_

3 days ago

It has begun! #NephJC #NSMC

**Nephrology Jrnl Club** @NephJC

3 days ago

@CharlieTomson @divyaa24 I concur! Welcome Charlie #NephJC

**Tom Oates @toates_19**

3 days ago

@NephJC Really? Flagging uncatalogued variants is the role of clinicians? Not sure that's true #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

@toates_19 @CharlieTomson @divyaa24 Must be a #NephJC record

**Nephrology Jrnl Club @NephJC**

3 days ago

T0: For some background on WGS check out this video <https://t.co/JWVhc7PABy> #NephJC

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@ClaraDay13 I HEART THIS POEM #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

TO: Additionally, check out the #NephMadness 2015: Genetic Nephrology Region, particularly the segment on NGS written and edited by Paul Phelan (@paulphel) and Conall O'Seaghda (@conall100) <https://t.co/vTKHD41Pfj> #NephJC

**Sarah Gleeson @sarah_gleeson_**

3 days ago

Hello everyone, Sarah here, renal trainee. Looking forward to #NephJC. No COI...slightly late

**Nephrology Jrnl Club @NephJC**

3 days ago

T0: 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

**Cathy Quinlan @KidneyCathy**

3 days ago

RT @KidneyCathy: Was there any way to feed back these results to the AURORA cohort? Esp for the AD conditions in patients who may have chil...

**Tess Harris @Elektra**

3 days ago

Tess. London. Popping in and out. No COI #NephJC



Dr MK #NephrologistOnICU @_Dr_MK

3 days ago

@DRuthclaire @RoyalFreeNHS BIG UP the @RoyalFreeNHS crew representing @NephJC - it's a wonder - we are meeting at #NephJC rather than in our hospital...



Nephrology Jrnl Club @NephJC

3 days ago

T0: These technologies may be particularly useful in CKD, given that it can go undetected until an advanced disease stage; at which point renal biopsy may be contraindicated or yield inconclusive results. #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T0: This lack of diagnostic certainty can impede clinical management, including tailored interventions #NephJC



Alexander Kirsch @alehkir

3 days ago

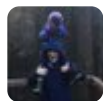
Alex, Graz, Austria; no COI #NephJC



Alex Hamilton @_ajhamilton

3 days ago

@NephJC We need a kidney gene panel we can run in these cases... #NephJC



Mark Jesky @MarkJesky

3 days ago

@NephJC Key here may be ability to 'alter patient management' and higher pick up rate #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@alehkir Welcome Alex #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@Elektra Welcome Tess #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

RT @_ajhamilton: @NephJC We need a kidney gene panel we can run in these cases... #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@sarah_gleeson_ Welcome Sarah #NephJC



Matt Bottomley @bottomleymatt

3 days ago

@NephJC Has anyone any idea of how much exome sequencing costs, for a single patient? #NephJC



Paul Phelan @paulphel

3 days ago

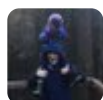
Hi all. Paul Phelan, Edinburgh. I'll be in and out. Great topic/study. No COI #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T0: In adults, exome sequencing has been applied in patients with FSGS. Check out this VA by @rheault_m @cjasn #NephJC <https://t.co/McyvTtDmgB>



Mark Jesky @MarkJesky

3 days ago

@NephJC Perhaps being overly cynical, but is lack of diagnostic certainty more troubling for patients or clinicians?? #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@MarkJesky Agreed. May being the operative word #NephJC



Clara Day @ClaraDay13

3 days ago

@bottomleymatt @NephJC Early doors for a cost- benefit question. Usually my job #nephjc



Nephrology Jrnl Club @NephJC

3 days ago

@paulphel Welcome Paul! #NephJC



Alex Hamilton @_ajhamilton

3 days ago

@MarkJesky @NephJC like @rheault_m says in her commentary, diagnosis hugely helpful for patients and family #NephJC



Vanessa Lerma @VanessaLerma17

3 days ago

Vanessa Lerma from Mexico, Nephrology resident No COI #NephJC #NSMC

**Nephrology Jrnl Club @NephJC**

3 days ago

T0: What percentage of your ERSD patients (roughly!) are labelled with "unknown aetiology"? How does this effect your clinical management? How do you outline this to the patient? #NephJC

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@DRuthclaire @RoyalFreeNHS @NephJC @marksandspencer Done and done!! I'm on the ICU @RoyalFreeNHS - let me know whenever you're around! #NephJC

**Paul Phelan @paulphel**

3 days ago

@bottomleymatt @NephJC Can a get a whole genome done in Scotland for £800. The work starts there though (basic QC, variant calling only) #NephJC

**Tom Oates @toates_19**

3 days ago

@MarkJesky @NephJC A fair qn, but @GenomicsEngland will tell you that one of the things 100k genomes proj has shown is how important ppl, especially parents, find a label of an otherwise mystery dz #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

RT @paulphel: @bottomleymatt @NephJC Can a get a whole genome done in Scotland for £800. The work starts there though (basic QC, variant ca...

**Charlie Tomson @CharlieTomson**

3 days ago

this commentary by @rheault_m is brilliant and changed my opinion. #nephjc

**Tom Oates @toates_19**

3 days ago

@paulphel @bottomleymatt @NephJC Exactly "cost of sequencing" isn't the issue imo #NephJC

**Sinead Stoneman @Stones__**

3 days ago

RT @toates_19: @MarkJesky @NephJC A fair qn, but @GenomicsEngland will tell you that one of the things 100k genomes proj has shown is how i...

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@ClaraDay13 @bottomleymatt @NephJC Is the spare change from #RenalBrexitt rattling in the tin?? #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



@MarkJesky A fair question. Doctors (NB nephrologists) hate diagnostic uncertainty. For patients, apart from their own prognosis, I think it is the implications for their families that often troubles them the most #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



@VanessaLerma17 Welcome Vanessa #NephJC

Tom Oates @toates_19

3 days ago



@MarkJesky @NephJC @GenomicsEngland Which of course also came across in @rheault_m's commentary for this #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



And now...T1 and the methods #NephJC.

Joel Topf, MD FACP @kidney_boy

3 days ago



RT @CharlieTomson: this commentary by @rheault_m is brilliant and changed my opinion. #nephjc <https://t.co/r1WW5yz6yr>

Sinead Stoneman @Stones__

3 days ago



RT @CharlieTomson: this commentary by @rheault_m is brilliant and changed my opinion. #nephjc <https://t.co/r1WW5yz6yr>

Paul Phelan @paulphel

3 days ago



@NephJC TO lots unknown and a further chuck mis-classified (a recent HNF1B diagnosis who thought they had diabetic nephropathy comes to mind), as this paper illustrates #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



T1: Proband only exome sequencing was performed on 3315 pts from 2 independent cohorts. First 1128 were from the AURORA cohort which included patients aged 50-80 and all-cause ESKD on HD >3 months. <https://t.co/3tG5sJazxf> #NephJC

Alex Hamilton @_ajhamilton

3 days ago



@toates_19 @MarkJesky @NephJC @GenomicsEngland @rheault_m snap #NephJC

**Mark Jesky** @MarkJesky

3 days ago

@CharlieTomson @rheault_m Agree commentary fantastic #nephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

@_Dr_MK @ClaraDay13 @bottomleymatt Will the dreaded B word affect our research and availability of sequencing technology? #NephJC #RenalBrexite

**Alexander Kirsch** @alehkir

3 days ago

@_ajhamilton @MarkJesky @NephJC @rheault_m Question is in what % mutation = diagnosis? #NephJC

**Louise Oni** @louise_oni

3 days ago

@NephJC sorry I'm late ☐ Paeds neph in UK #NephJC

**Rachel Lennon** @RLWczyk

3 days ago

Hello! Late but finally made #NephJC - paediatric nephrologist/basement membrane researcher Manchester no COI

**Tom Oates** @toates_19

3 days ago

@NephJC @_Dr_MK @ClaraDay13 @bottomleymatt Shouldn't do because of @GenomicsEngland #NephJC

**Sarah Gleeson** @sarah_gleeson_

3 days ago

@NephJC welcome! #NephJC

**Alex Hamilton** @_ajhamilton

3 days ago

@NephJC @_Dr_MK @ClaraDay13 @bottomleymatt 🔥 #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

T1: The remaining 2187 were from CUMC genetic study of CKD cohort which included patients with renal failure requiring dialysis or transplantation, serum creatinine > 132umol/L (>114umol/L in women] or the presence of severe proteinuria or hematuria. #NephJC



Sarah Gleeson @sarah_gleeson_
@ericau @NephJC Hi Eric! #NephJC

3 days ago



Matt Bottomley @bottomleymatt
@toates_19 @paulphel @NephJC Agreed, but it might limit use to select cases were it
£+++ #nephjc

3 days ago



Dr MK #NephrologistOnICU @_Dr_MK
@toates_19 @NephJC @ClaraDay13 @bottomleymatt @GenomicsEngland #sarcasm
@toates_19? #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
T1: Genomic DNA from participants (mostly peripheral blood) sequences were
captured and underwent paired-end sequencing for a target coverage of 111x at
Columbia Institute for genomic medicine. #NephJC

3 days ago



Sarah Gleeson @sarah_gleeson_
@RLWczyk Welcome! #NephJC

3 days ago



Tom Oates @toates_19
@RLWczyk Professor Lennon! #NephJC <https://t.co/GrCYcqKiJf>

3 days ago



Dr MK #NephrologistOnICU @_Dr_MK
@RLWczyk WOOHOO! WELCOME @RLWczyk to #NephJC

3 days ago



Sinead Stoneman @Stones__
RT @alehkir: @_ajhamilton @MarkJesky @NephJC @rheault_m Question is in what
% mutation = diagnosis? #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
T1: The investigators then focused their analyses on 625 genes they manually curated
associated with Mendelian forms of genitourinary disease #NephJC

3 days ago



Alex Hamilton @_ajhamilton
@NephJC You what now? #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
@ClaraDay13 Welcome Clara! #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
T1: Diagnostic variants were those that were classified as “pathogenic” or “likely pathogenic” according to the American College of Medical Genetics and Genomics (ACMG) guidelines <https://t.co/hb3U2jARud> #NephJC

3 days ago



Clara Day @ClaraDay13
@toates_19 @NephJC @_Dr_MK @bottomleymatt @GenomicsEngland Will be able to get genetic testing. Just no dialysers #nephjc 😊

3 days ago



Dr MK #NephrologistOnICU @_Dr_MK
@toates_19 @NephJC @ClaraDay13 @bottomleymatt @GenomicsEngland of course it will and it already has. Massive impact on research, universities, scientists. 100K Genome project only provides the raw data, not the people to do research on it? #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
T1: Due to different consent protocols, actionable genes in the CUMC patients sequence data s could be further interrogated and were separately analysed for pathogenic variants in the 59 genes recommended by the ACMG #NephJC

3 days ago



Nephrology Jrnl Club @NephJC
@alehkir @_ajhamilton @MarkJesky @rheault_m And that is the critical question facing it's applicability I would argue #NephJC

3 days ago



Charlie Tomson @CharlieTomson
@NephJC The @UKRenalRegistry report some years ago showed marked variation in the use of the 'uncertain' code (00) for primary renal disease - balanced by more use of codes for equally un-proveable diagnoses like 'hypertensive nephropathy'. Searching for the reference....#nephjc

3 days ago



Clara Day @ClaraDay13
@toates_19 @_Dr_MK @NephJC @bottomleymatt @GenomicsEngland If only £800 probably better value than 6 months phosphate binders #nephjc

3 days ago

**Nephrology Jrnl Club** @NephJC

3 days ago

T1: Any thoughts about the methods? #NephJC

**Dr MK #NephrologistOnICU** @_Dr_MK

3 days ago

@ClaraDay13 @toates_19 @NephJC @bottomleymatt @GenomicsEngland 🤔🤔🤔
#NephJC**Tom Oates** @toates_19

3 days ago

@ClaraDay13 @NephJC @_Dr_MK @bottomleymatt @GenomicsEngland Don't recall anything about dialysers in the #TopolReview lol @MattHancock #dothepresent
#NephJC**Damian Fogarty** @DamianFog

3 days ago

Tweet chat started on this fascinating study #NephJC. I'm on flight back home after #ABCDRenal19 so this is my sole contribution!

**Rob Peel** @rabpeel

3 days ago

@ClaraDay13 @toates_19 @_Dr_MK @NephJC @bottomleymatt
@GenomicsEngland I agree #nephjc**Sinead Stoneman** @Stones__

3 days ago

RT @CharlieTomson: @NephJC The @UKRenalRegistry report some years ago showed marked variation in the use of the 'uncertain' code (00) for p...

**Nephrology Jrnl Club** @NephJC

3 days ago

RT @CharlieTomson: this commentary by @rheault_m is brilliant and changed my opinion. #nephjc <https://t.co/r1WW5yz6yr>**Clara Day** @ClaraDay13

3 days ago

@toates_19 @NephJC @_Dr_MK @bottomleymatt @GenomicsEngland
@MattHancock Sorry #nephjc And there will be dialysers honest**Nephrology Jrnl Club** @NephJC

3 days ago

RT @CharlieTomson: @NephJC The @UKRenalRegistry report some years ago showed marked variation in the use of the 'uncertain' code (00) for p...



Tom Oates @toates_19

3 days ago

@ClaraDay13 @_Dr_MK @NephJC @bottomleymatt @GenomicsEngland Dr Day on proper lol form tonight I see #NephJC <https://t.co/PrMDWXCydl>



Nephrology Jrnl Club @NephJC

3 days ago

@louise_oni Welcome Louise, good to have you along, no apologies needed #NephJC



Alexander Kirsch @alehkir

3 days ago

@NephJC Total ignorant on this topic: is there enough data to be sure that the other variants are not pathogenic or is this a non-exclusive list? #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@RLWczyk Welcome @RLWczyk to #NephJC!



Donal Reddan @DREDD001

3 days ago

#NephJC Donal Reddan Galway . No COI following with interest



Nephrology Jrnl Club @NephJC

3 days ago

On to T2 and the results. #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T2: The cohort was predominantly adult (91.6% were >21 years of age), and 1179 patients (35.6%) reported having non-European ancestry #NephJC <https://t.co/eFWckDQPWZ>



Daniyal Jafree @daniyal_jafree

3 days ago


@NephJC finally here after an expectedly late finish in the lab. Lurking but looking forward to reading back over people's thoughts on this one! No COI #NephJC #NSMC @NSMCInternship





Matt Bottomley @bottomleymatt

3 days ago


@NephJC @alehkir @_ajhamilton @MarkJesky @rheault_m Surely the gold standard at present is a renal biopsy (though I see a lot showed FSGS). It's not clear to me how the 'clinical diagnosis' was made in this paper - what proportion had a biopsy vs clinical judgement re: the diagnosis? #nephjc

 **Nephrology Jrnl Club @NephJC** 3 days ago
T2: Of note, the CUMC cohort included younger adults than AURORA, was more ethnically diverse and had a disproportionate number of glomerular patients #NephJC


 **Nephrology Jrnl Club @NephJC** 3 days ago
@DREDD001 Welcome @DREDD001 to #NephJC,


 **Dr MK #NephrologistOnICU @_Dr_MK** 3 days ago
@alehkir @NephJC absolutely not exclusive - exome sequencing = cherry picking in the orchard of genetic information #NephJC


 **Nephrology Jrnl Club @NephJC** 3 days ago
@daniyal_jafree @NSMCInternship Welcome @daniyal_jafree. Lurkers welcome! #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
RT @bottomleymatt: @NephJC @alehkir @_ajhamilton @MarkJesky @rheault_m Surely the gold standard at present is a renal biopsy (though I see...

 **Sarah Gleeson @sarah_gleeson_** 3 days ago
@daniyal_jafree @NephJC @NSMCInternship welcome! #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
T2: The cohort included a wide breadth of clinical diagnosis including nephropathy of unknown origin (8.5%). 64.7% had ESKD. 28.3% had a positive family history of kidney disease #NephJC

 **Paul Phelan @paulphel** 3 days ago
@alehkir @NephJC There's likely novel genes outside of this list in these patients but would need to interrogate other affected family members I guess to find them. More work from this group I hope. #nephjc

 **Sinead Stoneman @Stones__** 3 days ago
@bottomleymatt @NephJC @alehkir @_ajhamilton @MarkJesky @rheault_m Good point. Particularly for 'hypertensive nephropathy' types etc. #nephjc



Sinead Stoneman @Stones__

3 days ago

RT @bottomleymatt: @NephJC @alehkir @_ajhamilton @MarkJesky @rheault_m Surely the gold standard at present is a renal biopsy (though I see...



Nephrology Jrnl Club @NephJC

3 days ago

T2: Diagnostic variants were detected in 9.3% of the entire cohort (307 patients) spanning 66 different 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/VhhkVB0tU7>



Nephrology Jrnl Club @NephJC

3 days ago

@bottomleymatt @alehkir @_ajhamilton @MarkJesky @rheault_m I can't find mention in the paper or supplementary material though I stand to be corrected. Excellent point #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T2: Six diseases accounted for 63% of the genetic diagnoses, with mutations in PKD1 affecting the largest number of patients. #NephJC <https://t.co/49D6REbSI1>



Matt Graham-Brown @DrMattGB

3 days ago

MGB mega late. Apologies. Observing. Genetics novice no COI #NephJC



Alex Hamilton @_ajhamilton

3 days ago

@DrMattGB I love that you've become your own acronym #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T2: 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/SYudo01ZMO>



Louise Oni @louise_oni

3 days ago

@NephJC #NephJC sorry forgot #



Paul Phelan @paulphel

3 days ago

@NephJC Interesting there was a handful of PKD1 with 'unknown' phenotype #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



T2: Examining this data further, it appears the diagnostic yield for nephropathy or unknown origin is higher in the AURORA cohort. There was a high predictive diagnostic yield for glomerulopathy in the AURORA cohort-more representative of community sample #NephJC

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@louise_oni @NephJC I second this #NephJC

**Rob Peel @rabpeel**

3 days ago

@_ajhamilton @DrMattGB if he was an MGB GT perhaps he would be on time :-)
#nephjc

Nephrology Jrnl Club @NephJC

3 days ago



T2: There was a lower diagnostic yield for glomerulopathy in the CUMC cohort (likely related to local expertise). #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

@DrMattGB Welcome MGB to #NephJC (rhyming acronym)

**Nephrology Jrnl Club @NephJC**

3 days ago

@louise_oni Agreed. #NephJC

**Matt Graham-Brown @DrMattGB**

3 days ago

@rabpeel @_ajhamilton Lol. It's like being back at school #nephjc
<https://t.co/RqBV2JMIUI>

**Renal Association @RenalAssoc**


3 days ago


Nephrology learning double header - catch up on #ABCDRenal19 from earlier today as #NephJC unfolds in real time


**Nephrology Jrnl Club @NephJC**


3 days ago


T2: A disease subtype was identified in 65 patients and disease was reclassified in 18 patients which may aid more precise management #NephJC


 **Nephrology Jrnl Club @NephJC** 3 days ago
T2: For 89% of the CUMC patients with genetic findings from the exome sequencing, the molecular diagnosis has implications to alter their clinical management. #NephJC
<https://t.co/owgFKNRus7>


 **Sarah Gleeson @sarah_gleeson_** 3 days ago
RT @RenalAssoc: Nephrology learning double header - catch up on #ABCDRenal19 from earlier today as #NephJC unfolds in real time


 **Hugo Diniz @HDiniz_** 3 days ago
RT @NephJC: T2: For 89% of the CUMC patients with genetic findings from the exome sequencing, the molecular diagnosis has implications to a...


 **Nephrology Jrnl Club @NephJC** 3 days ago
T2: 39 CUMC patients with “nephropathy of unknown origin” had a molecular cause of their CKD identified. #NephJC

 **Daniyal Jafree @daniyal_jafree** 3 days ago
@NephJC @NSMCInternship Thanks, my excuse is that I’m working towards my first kidney paper. Wondering if it could potentially make the cut for a future #NephJC (!?) 🙄

 **Alex Hamilton @_ajhamilton** 3 days ago
@NephJC We haven’t touched on the value of genetic diagnosis when working up potential family LKD donors yet #NephJC

 **Matt Graham-Brown @DrMattGB** 3 days ago
I don’t know much, but I expect this is true #nephjc

 **Nephrology Jrnl Club @NephJC** 3 days ago
@toates_19 Applicable to the research end only....I hope #NephJC

 **Nephrology Jrnl Club @NephJC** 3 days ago
RT @RenalAssoc: Nephrology learning double header - catch up on #ABCDRenal19 from earlier today as #NephJC unfolds in real time



Nephrology Jrnl Club @NephJC

3 days ago

T2: 62% of patients with COL4A3, COL4A4, or COL4A5 mutations did not have clinical diagnoses of Alport syndrome or thin basement membrane disease. This would merit ophthalmologic and otolaryngologic referral #NephJC



Sinead Stoneman @Stones__

3 days ago

@paulphel @NephJC Agree. Reminds me of the NPHP1 paper (<https://t.co/fb9CuBZ3DB>). Is this variable expression due to disease modifying genes or just that when these diseases were described the classic presentation predated the genetic info we are now discovering. #NephJC



Louise Oni @louise_oni

3 days ago

RT @DrMattGB: I don't know much, but I expect this is true #nephjc
<https://t.co/w6SpXeNeeh>



Nephrology Jrnl Club @NephJC

3 days ago

RT @_ajhamilton: @NephJC We haven't touched on the value of genetic diagnosis when working up potential family LKD donors yet #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

@_ajhamilton Could indeed be of huge value #NephJC



Louise Oni @louise_oni

3 days ago

@_ajhamilton @NephJC This is v important #NephJC the last thing you want is the donor ending up in ESRF



Nephrology Jrnl Club @NephJC

3 days ago

T2: Pathogenic variants were detected in 16 of the 59 ACMG actionable genes in 1.6% (34/2197) patients in the CUMC cohort. #NephJC



Alex Hamilton @_ajhamilton

3 days ago

@louise_oni @NephJC whoops #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

RT @paulphel: @NephJC Interesting there was a handful of PKD1 with 'unknown' phenotype #NephJC



Sarah Gleeson @sarah_gleeson_

3 days ago

RT @_ajhamilton: @NephJC We haven't touched on the value of genetic diagnosis when working up potential family LKD donors yet #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T2: These secondary findings may also have implications for nephrology care including immunosuppression in patients with hereditary malignancy, diuretic prescription and predisposition to arrhythmia. #NephJC



Alex Hamilton @_ajhamilton

3 days ago

@NephJC Put the biopsy needle down! #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T2: Any final comments on the results? #NephJC



Rob Peel @rabpeel

3 days ago

@NephJC @_ajhamilton We have a familial IgA clan and one had donated in London before it came to light. not sure of genetics though #NephJC



Charlie Tomson @CharlieTomson

3 days ago

@NephJC @UKRenalRegistry found it. Marked centre-to-centre variation in primary disease coding, nearly certainly not driven by real variation but by clinician bias/variation in approach to ascertainment of PRD <https://t.co/YW9BViRR4H> #nephjc <https://t.co/DqyK6w88wz>



Rob Peel @rabpeel

3 days ago

@NephJC T2 hopeful #nephjc



Nephrology Jrnl Club @NephJC

3 days ago

T3 and Conclusions: This study concluded that exome sequencing in a combined cohort of more than 3000 patients with CKD yielded a genetic diagnosis in just under 10% of cases. #NephJC



Rachel Lennon @RLWczyk

3 days ago

@NephJC Wonder how many had biopsies/careful EM analysis of GBM thickness? #NephJC

**Rob Peel @rabpeel**

3 days ago

T2 hopefully a higher yield of diagnosis as time goes on #nephjc

**Paul Phelan @paulphel**

3 days ago

@_ajhamilton @NephJC Absolutely. Probably the most common reason for genetic referral (& general hereditary worry) in my practice. We're v limited in what we can test for in NHS currently. It will change however #NephJC

**Matt Graham-Brown @DrMattGB**

3 days ago

@RLWczyk @NephJC Excellent point. #NephJC

**Sinead Stoneman @Stones__**

3 days ago

RT @CharlieTomson: @NephJC @UKRenalRegistry found it. Marked centre-to-centre variation in primary disease coding, nearly certainly not dri...

**Nephrology Jrnl Club @NephJC**

3 days ago

T3: Some limitations of exome sequencing as noted by the authors #NephJC

**Cathy Quinlan @KidneyCathy**

3 days ago

RT @ClaraDay13: @toates_19 @_Dr_MK @NephJC @bottomleymatt @GenomicsEngland If only £800 probably better value than 6 months phosphate binde...

**Nephrology Jrnl Club @NephJC**

3 days ago

T3: •Suboptimal coverage of the mitochondrial genome or the duplicated regions of PKD1 •Inability to detect intronic and copy-number variants •Likely underestimates the overall burden of genetic disorders among patients with nephropathy #NephJC

**Mark Jesky @MarkJesky**

3 days ago

@CharlieTomson @NephJC @UKRenalRegistry That's really interesting! #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

RT @CharlieTomson: @NephJC @UKRenalRegistry found it. Marked centre-to-centre variation in primary disease coding, nearly certainly not dri...



Cathy Quinlan @KidneyCathy

3 days ago

@NephJC @_Dr_MK @ClaraDay13 @bottomleymatt But under EU legislation the #AURORA participants in this study can access their results. After #brexit they can't #NephJc



Louise Oni @louise_oni

3 days ago

@DrMattGB @RLWczyk @NephJC Yep I agree, sounds suspicious & histological analysis open to observer errors #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T3: Would be interested in the correlation with kidney biopsy as opposed to clinical diagnosis alone #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T3: This study serves to emphasise the high degree of genetic and phenotypic heterogeneity of clinical disease in CKD.#NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T3: Genetic testing must be interpreted in the clinical context including clinical history and is unlikely to replace renal biopsy completely e.g. in the instance of evaluating degree of fibrosis #NephJC



Hugo Diniz @HDiniz_

3 days ago

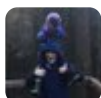
@NephJC What are the clinical implications of the first two? #NephJC



Nephrology Jrnl Club @NephJC

3 days ago

T3: Additionally, is there a risk of falsely attributing causality in few patients where genetic variants are not well specified? #NephJC



Mark Jesky @MarkJesky

3 days ago

@rabpeel And getting better at *choosing* the right people to do the test on in the first place #nephJC



Nephrology Jrnl Club @NephJC

3 days ago

T3: Any other limitations? #NephJC

Paul Phelan @paulphel

3 days ago



@NephJC Absolutely will be more genetic disease in there. Despite lots of PKD1&2 who were mostly known, I think the hit rate here was pretty high considering it's an adult, all comer CKD cohort, with mostly no family history #NephJC

Matt Bottomley @bottomleymatt

3 days ago



@NephJC Definitely #NephJC

Cathy Quinlan @KidneyCathy

3 days ago



@NephJC I think particularly so when you can't go back to patient details to verify phenotype. In this paper the CUMC V AURORA cohorts #NephJC

Alex Hamilton @_ajhamilton

3 days ago



Don't have a feel for how many of these monogenic diseases might occur de novo. #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



T3: A genetic diagnosis for patients, particularly those with "CKD aetiology unknown" is highly attractive for patient and physician; it enables us to attempt to predict and screen for complications and hopefully allow targeted treatment based on the underlying disease #NephJC

Hugo Diniz @HDiniz_

3 days ago



@_ajhamilton I also questioned myself about this. I don't think there is detailed information about that in the paper #nephjc

Nephrology Jrnl Club @NephJC

3 days ago



T3: In addition to the patient benefit, genetic diagnosis has significant potential in the workup of living kidney donors in terms of donor risk #NephJC

Nephrology Jrnl Club @NephJC

3 days ago



T3: Any other thoughts on benefits? #NephJC



Charlie Tomson @CharlieTomson

3 days ago

@RLWczyk @NephJC Do we know whether patients with a COL4 mutation who don't have 'classical' Alport syndrome derive the same benefit from early initiation of ACEI Rx as do those with classical biopsy findings? #nephjc Is it the genotype of the phenotype that determines treatment response?



Rob Peel @rabpeel

3 days ago

T3 early days, when to test, what to test for and how to interpret it and all at an affordable price a few years away yet. #nephjc



Charlie Tomson @CharlieTomson

3 days ago

RT @NephJC: T3: A genetic diagnosis for patients, particularly those with “CKD aetiology unknown” is highly attractive for patient and phys...



Sinead Stoneman @Stones__

3 days ago

@MarkJesky @CharlieTomson @NephJC @UKRenalRegistry Reflects different levels of tolerance for diagnostic uncertainty and also dictated by clinical/patient circumstances to a degree too I imagine. #nephjc



Nephrology Jrnl Club @NephJC

3 days ago

How do you envisage these results will change your clinical practice? Crystal ball may be required #NephJC



Mark Jesky @MarkJesky

3 days ago

@NephJC Do people think including PKD1 and PKD2 inflates diagnostic yield and utility of exome sequencing? We use more targeted genetic test. #nephjc



Nephrology Jrnl Club @NephJC

3 days ago

RT @rabpeel: T3 early days, when to test, what to test for and how to interpret it and all at an affordable price a few years away yet. #ne...



Hugo Diniz @HDiniz_

3 days ago

@rabpeel How few? :P #NephJC

**Sinead Stoneman @Stones__**

3 days ago

RT @NephJC: T3: This study serves to emphasise the high degree of genetic and phenotypic heterogeneity of clinical disease in CKD.#NephJC

**Charlie Tomson @CharlieTomson**

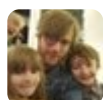
3 days ago

@NephJC All true, but we're desperately in need of more 'targeted treatments' for well-defined underlying disease, whether genetically determined or not - there really aren't many such treatments out there - yet... #nephjc

**Sinead Stoneman @Stones__**

3 days ago

RT @NephJC: T3: Genetic testing must be interpreted in the clinical context including clinical history and is unlikely to replace renal bio...

**Dean Wallace @DrDeano**

3 days ago

#nephjc super late to the party, Dean Wallace, UK, Paeds Neph. Long time listener

**Louise Oni @louise_oni**

3 days ago

@NephJC #NephJC as above sorry

**Rob Peel @rabpeel**

3 days ago

@HDiniz_ 5-10 years at a guess? #NephJC

**Alex Hamilton @_ajhamilton**

3 days ago

@NephJC Think genetic risk scores are useful to help justify cost of testing #NephJC

**Sinead Stoneman @Stones__**

3 days ago

RT @_ajhamilton: Don't have a feel for how many of these monogenic diseases might occur de novo. #NephJC

**Nephrology Jrnl Club @NephJC**

3 days ago

@CharlieTomson Agreed, unfortunately I feel like we've been hearing "target treatments" on the horizon for quite some time #NephJC



Sinead Stoneman @Stones__

3 days ago

RT @paulphel: @NephJC Absolutely will be more genetic disease in there. Despite lots of PKD1&2 who were mostly known, I think the hit rate...



Rachel Lennon @RLWczyk

3 days ago

@_ajhamilton Around 15% de novo in X-linked Alport syndrome referenced in: <https://t.co/2IMvGv1Bai> #NephJC



Clara Day @ClaraDay13

3 days ago

Actually I went I. To this chat thinking *why bother testing* and I think my mind might have been changed. Nhs e testing is now much easier but just have to use selectively. Unlike phos binders usage #nephjc



Nephrology Jrnl Club @NephJC

3 days ago

Final few minutes; Take-home points? #NephJC



Sinead Stoneman @Stones__

3 days ago

RT @NephJC: T3: Additionally, is there a risk of falsely attributing causality in few patients where genetic variants are not well specifie...



Nephrology Jrnl Club @NephJC

3 days ago

@DrDeano Welcome Dean! #NephJC



Sinead Stoneman @Stones__

3 days ago

RT @CharlieTomson: @RLWczyk @NephJC Do we know whether patients with a COL4 mutation who don't have 'classical' Alport syndrome derive the...



Nephrology Jrnl Club @NephJC

3 days ago

Thanks for joining us tonight and for contributing to the discussion. #NephJC



Paul Phelan @paulphel

3 days ago

T3 WES/WGS will become an available test for CKD workup. It'll be cheaper (already affordable) and as we sequence more and more humans our ability to call pathogenic variants will exponentially rise #nephjc

**Hugo Diniz** @HDiniz_

3 days ago

@rabpeel Yeah, 5 years seems a good guess. They are already cheap enough to use in a clinical setting, and will probably get cheaper as time goes by. #NephJC

**Sinead Stoneman** @Stones__

3 days ago

RT @rabpeel: T3 early days, when to test, what to test for and how to interpret it and all at an affordable price a few years away yet. #ne...

**Dr MK #NephrologistOnICU** @_Dr_MK

3 days ago

@ClaraDay13 This is an important point and a good 'take-home' #NephJC

**Alex Hamilton** @_ajhamilton

3 days ago

@RLWczyk Useful to remember that >1/10 may not have family history! #NephJC

**Tess Harris** @Elektra

3 days ago

RT @ClaraDay13: Actually I went I. To this chat thinking *why bother testing* and I think my mind might have been changed. Nhs e testing is...

**Matt Bottomley** @bottomleymatt

3 days ago

@Stones__ @MarkJesky @CharlieTomson @NephJC @UKRenalRegistry Agreed. Have worked in two centres - one where they had capacity to do 2-3 biopsies per week; other where they were doing 2-3 per day. Guess in which centre people tended to get a biopsy? #nephjc

**Tess Harris** @Elektra

3 days ago

RT @_ajhamilton: @NephJC Think genetic risk scores are useful to help justify cost of testing #NephJC

**Matt Graham-Brown** @DrMattGB

3 days ago

@CharlieTomson @RLWczyk @NephJC Don't know. I expect most clinicians would be more likely to suggest ACE/ARB in phenotypically atypical X-linked and AR Alport's than AD Alport's, but no empirical data behind that, just a hunch. #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

RT @ClaraDay13: Actually I went I. To this chat thinking *why bother testing* and I think my mind might have been changed. Nhs e testing is...

**Nephrology Jrnl Club @NephJC**

3 days ago

Thanks again to @yasar_caliskan and @jenniejlin for the summary, @rheault_m for the commentary and @divyaa24 for the VA. #NephJC

**Charlie Tomson @CharlieTomson**

3 days ago

@Stones__ @MarkJesky @NephJC @UKRenalRegistry agree - and also whether 'hypertensive nephropathy' is a 'real' diagnosis <https://t.co/yNGvt8yLZ0> #nephjc

**Alexander Kirsch @alehkir**

3 days ago

Wondering how soon a similar test will be commercialized, even direct to consumer (patient). We need to be aware of these developments. How to react and advise, when this is done and we are confronted with genetic testing we never ordered. #NephJC

**Matt Bottomley @bottomleymatt**

3 days ago

@NephJC I think it'll be useful in select cases, where lack of diagnosis is causing significant distress or whether LRD is being considered. Would need significant counselling about limitations at present though. #nephjc

**Tess Harris @Elektra**

3 days ago

RT @NephJC: T3: In addition to the patient benefit, genetic diagnosis has significant potential in the workup of living kidney donors in te...

**Nephrology Jrnl Club @NephJC**

3 days ago

Please also sign up for our weekly newsletter at <https://t.co/5aumn8qL85> #NephJC

**Hugo Diniz @HDiniz_**

3 days ago

RT @paulphel: T3 WES/WGS will become an available test for CKD workup. It'll be cheaper (already affordable) and as we sequence more and mo...

**Nephrology Jrnl Club @NephJC**

3 days ago

RT @bottomleymatt: @NephJC I think it'll be useful in select cases, where lack of diagnosis is causing significant distress or whether LRD...

**Nephrology Jrnl Club @NephJC**

3 days ago

RT @CharlieTomson: @Stones__ @MarkJesky @NephJC @UKRenalRegistry agree - and also whether 'hypertensive nephropathy' is a 'real' diagnosis...



Hugo Diniz @HDiniz_

3 days ago

@paulphel I think this is a great conclusion! For CKD this paper is probably the beginning of it #nephjc



Swapnil Hiremath, MD, MPH @hswapnil

3 days ago

RT @paulphel: T3 WES/WGS will become an available test for CKD workup. It'll be cheaper (already affordable) and as we sequence more and mo...



Dr MK #NephrologistOnICU @_Dr_MK

3 days ago

@NephJC I think we need to move away from the "why test when the treatment isn't different" argument to the debate we have whenever discussing all other #NephJC research when we bemoan the heterogeneity of our clinical population - we can only develop treatment if we define ds accurately



Sinead Stoneman @Stones__

3 days ago

RT @CharlieTomson: @Stones__ @MarkJesky @NephJC @UKRenalRegistry agree - and also whether 'hypertensive nephropathy' is a 'real' diagnosis...



Swapnil Hiremath, MD, MPH @hswapnil

3 days ago

#NephJC



Nephrology Jrnl Club @NephJC

3 days ago

Looking for some #NephJC swag? Order it here <https://t.co/eTUZY27Qo>



Nephrology Jrnl Club @NephJC

3 days ago

RT @paulphel: T3 WES/WGS will become an available test for CKD workup. It'll be cheaper (already affordable) and as we sequence more and mo...



Tess Harris @Elektra

3 days ago

RT @_ajhamilton: @NephJC We haven't touched on the value of genetic diagnosis when working up potential family LKD donors yet #NephJC



Paul Phelan @paulphel

3 days ago

@CharlieTomson @Stones__ @MarkJesky @NephJC @UKRenalRegistry yes I bet it almost never is. APOL1 has debunked most hypertensive ESKD in AA's.... #NephJC

**Tess Harris @Elektra**

3 days ago

RT @paulphel: @_ajhamilton @NephJC Absolutely. Probably the most common reason for genetic referral (& general hereditary worry) in my prac...

**Mark Jesky @MarkJesky**

3 days ago

@bottomleymatt @Stones__ @CharlieTomson @NephJC @UKRenalRegistry I am sure there must be a intra (and inter) centre variation of performing renal biopsy - not sure if it's been looked at though.... #nephJc

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

3 days ago

RT @CharlieTomson: @NephJC @UKRenalRegistry found it. Marked centre-to-centre variation in primary disease coding, nearly certainly not dri...

**Sinead Stoneman @Stones__**

3 days ago

RT @paulphel: @CharlieTomson @Stones__ @MarkJesky @NephJC @UKRenalRegistry yes I bet it almost never is. APOL1 has debunked most hypertensi...

**Matt Bottomley @bottomleymatt**

3 days ago

Thanks for hosting @NephJC! Thoroughly enjoyed my first #nephjc!

**Tess Harris @Elektra**

3 days ago

RT @alehkir: Wondering how soon a similar test will be commercialized, even direct to consumer (patient). We need to be aware of these deve...

**Nephrology Jrnl Club @NephJC**

3 days ago

Until next time...thank you and good night! #NephJC

**Tess Harris @Elektra**

3 days ago

RT @alehkir: Wondering how soon a similar test will be commercialized, even direct to consumer (patient). We need to be aware of these deve...

**Alex Hamilton @_ajhamilton**

3 days ago

@alehkir #NephJC <https://t.co/Beb7uMZ17u>



Cathy Quinlan @KidneyCathy

3 days ago

RT @CharlieTomson: @NephJC @UKRenalRegistry found it. Marked centre-to-centre variation in primary disease coding, nearly certainly not dri...



Sinead Stoneman @Stones__

3 days ago

RT @paulphel: T3 WES/WGS will become an available test for CKD workup. It'll be cheaper (already affordable) and as we sequence more and mo...



Matt Graham-Brown @DrMattGB

3 days ago

@_Dr_MK @NephJC Fair shout #nephjc



Cathy Quinlan @KidneyCathy

3 days ago

@bottomleymatt @Stones__ @MarkJesky @CharlieTomson @NephJC @UKRenalRegistry When you have a hammer. Everything looks like a nail. #nephjc <https://t.co/xmMPoNnNOY>



Clara Day @ClaraDay13

3 days ago

@KidneyCathy I am going through a phase of seeing lots of our s asian population with CKD with numbers of relatives on dialysis. They are scared. We have great genetic renal service and very helpful #nephjc



Alex Hamilton @_ajhamilton

3 days ago

RT @KidneyCathy: @bottomleymatt @Stones__ @MarkJesky @CharlieTomson @NephJC @UKRenalRegistry When you have a hammer. Everything looks like...



Tess Harris @Elektra

3 days ago


RT @bottomleymatt: @NephJC I think it'll be useful in select cases, where lack of diagnosis is causing significant distress or whether LRD...




Sinead Stoneman @Stones__

3 days ago


RT @bottomleymatt: @NephJC I think it'll be useful in select cases, where lack of diagnosis is causing significant distress or whether LRD...


 **Charlie Tomson @CharlieTomson** 3 days ago
#nephjc whole exome/whole genome sequencing likely to become a major part of the work-up of someone with kidney disease - will watch with fascination to see how this drives research into prevention/early treatment. Big implications for @Kidney_Research and other funders


 **Cathy Quinlan @KidneyCathy** 3 days ago
Brilliant point. #NephJc


 **Sinead Stoneman @Stones__** 3 days ago
RT @_Dr_MK: @NephJC I think we need to move away from the "why test when the treatment isn't different" argument to the debate we have when...


 **Alex Hamilton @_ajhamilton** 3 days ago
@MarkJesky @bottomleymatt @Stones__ @CharlieTomson @NephJC @UKRenalRegistry .@toates_19 tweeted about this recently... #NephJC

 **Hugo Diniz @HDiniz_** 3 days ago
Great discussion! This paper may as well be the dawn of a new era in the diagnosis and management of CKD! #NephJC #NSMC

 **Rob Peel @rabpeel** 3 days ago
thanks to everyone for my fix of science! #nephjc

 **Cathy Quinlan @KidneyCathy** 3 days ago
@NephJC Thanks for a stimulating start to my day. Another excellently moderated #NephJc

 **Sinead Stoneman @Stones__** 3 days ago
Thanks for a great job hosting @Slatts_1 #NephJC

 **Paul Phelan @paulphel** 3 days ago
@MarkJesky @NephJC WES actually not a great way of testing for PKD1 #NephJC

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@KidneyCathy @bottomleymatt @Stones__ @MarkJesky @CharlieTomson @NephJC @UKRenalRegistry Or alternatively: "Whosoever holds this hammer, if he be worthy, shall possess the power of THOR!" #NephJC

**Mark Jesky @MarkJesky**

3 days ago

@paulphel @NephJC Agree. Certainly done differently in our PKD clinic. #nephjc

**Matt Graham-Brown @DrMattGB**

3 days ago

@MarkJesky @bottomleymatt @Stones__ @CharlieTomson @NephJC @UKRenalRegistry Massive! Undoubtedly individuals will change their practice with time and experience too. Suspect clinicians become more selective as they gain experience.... I just want to biopsy everything! #nephjc

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

RT @KidneyCathy: Brilliant point. #NephJc <https://t.co/ad1GBtMVyQ>

**Nephrology Jrnl Club @NephJC**

3 days ago

RT @CharlieTomson: #nephjc whole exome/whole genome sequencing likely to become a major part of the work-up of someone with kidney disease...

**Alex Hamilton @_ajhamilton**

3 days ago

That was a brilliant #NephJC - thanks all

**Matt Graham-Brown @DrMattGB**

3 days ago

Well done @Slatts_1 hosting. Tough chat, expertly handled 👍👏👏 #nephjc

**Dr MK #NephrologistOnICU @_Dr_MK**

3 days ago

@NephJC Thank you, as always, for a superb #NephJC chin wag!

**Alex Hamilton @_ajhamilton**

3 days ago

@DrMattGB @Slatts_1 Agreed - top drawer #NephJC

**Alex Hamilton @_ajhamilton**

3 days ago

@ClaraDay13 @KidneyCathy Did you make a genetic diagnosis? #NephJC

**Tom Oates** @toates_19

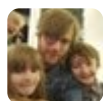
3 days ago

#PIVOTAL next on #NephJC by the way. Feb 26/7 #OneForTheClinicians

**Laura Slattery** @Slatts_1

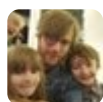
3 days ago

@_ajhamilton @DrMattGB Thanks @_ajhamilton and @DrMattGB, was a great discussion #NephJC

**Dean Wallace** @DrDeano

3 days ago

#NephJC I didn't have long enough to come up with any pithy insights. Great discussion to flick through though Thanks @NephJC

**Dean Wallace** @DrDeano

3 days ago

We do lots of targeted genetics in our practice mostly PAX2, uromodulin, HNF1B, NPHP for those pesky bright or cystic looking kidneys. CoL4 in haematuria/proteinuria #NephJC

**Dean Wallace** @DrDeano

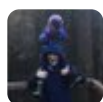
3 days ago

Bristol steroid resistant nephrotic screen even in those with unexplained heavy proteinuria without nephrosis #nephjc

**Tomas Rohal** @calvapo

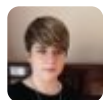
3 days ago

@NephJC Sorry to have missed you tonight, it looks like it was a nice JC. Hope to meet you next time #NephJC

**Mark Jesky** @MarkJesky

3 days ago

@DrMattGB @bottomleymatt @Stones__ @CharlieTomson @NephJC @UKRenalRegistry Being pro- biopsy certainly not bad, but need to justify procedure. Often depends on what patients and their families want - and whether potential transplant recipient - some individuals (patients and clinicians) can cope with the unknown unknowns better than others. #nephjc

**Laura Slattery** @Slatts_1

3 days ago

RT @toates_19: #PIVOTAL next on #NephJC by the way. Feb 26/7 #OneForTheClinicians

**Alex Hamilton** @_ajhamilton

3 days ago

This #NephJC chat has got me excited about #ukkw2019

**Clara Day** @ClaraDay13

3 days ago

@_ajhamilton @KidneyCathy Brewing...#nephjc

**Tom Oates** @toates_19

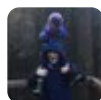
3 days ago

@DrDeano Deano Deano Deano #NephJC

**Mark Jesky** @MarkJesky

3 days ago

@_Dr_MK @KidneyCathy @bottomleymatt @Stones__ @CharlieTomson @NephJC @UKRenalRegistry I hope Thor doesn't send me a DOPS request with that biopsy technique #nephjc #stillrequiressupervison

**Mark Jesky** @MarkJesky

3 days ago

@ClaraDay13 @_ajhamilton @KidneyCathy Having a clinical genetics team interested in nephrology really key in these cases #nephJC

**Rachel Lennon** @RLWczyk

3 days ago

@NephJC Thank you! Loved #NephJC

**Nephrology Jrnl Club** @NephJC

3 days ago

RT @DrDeano: We do lots of targeted genetics in our practice mostly PAX2, uromodulin, HNF1B, NPHP for those pesky bright or cystic looking...

**Tom Oates** @toates_19

3 days ago

@RLWczyk @NephJC Professor Lennon! No wait, I've done that joke #NephJC <https://t.co/Zte9FGwhEh>**Cathy Quinlan** @KidneyCathy

3 days ago

@_ajhamilton @ClaraDay13 You need to work in Melbourne where we can get a 6month, 4week or 72hour turnaround. #nephjc @GenomeSeb

**Alex Hamilton** @_ajhamilton

3 days ago

@KidneyCathy @ClaraDay13 @GenomeSeb ☐ #nephjc



Cathy Quinlan @KidneyCathy

3 days ago

@MarkJesky @ClaraDay13 @_ajhamilton Definitely. Multidisciplinary approach raises the diagnostic rate. Watch this space... #nephjc




Joe Lafferty @thejoelafferty

3 days ago

RT @AdvDialysis: New study raises questions of whether the widely accepted standard of care in thrice-weekly #hemodialysis is simply inadeq...

Showing 1 to 333 of 333 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.
Get unrestricted access to all conversations with **Symplur Signals**.

©2019 Symplur LLC – Connecting the dots in healthcare social media