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# RACP Foundation Research Awards

# FINAL REPORT

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| **Project / Program Title** | | Genetic Diagnostics in Genetic Renal Disease - Methods, Applications and Therapeutics |
| **Name** | | Dr Andrew John Mallett |
| **Award Received** | | 2015 Jacquot Research Entry Scholarships in Nephrology |
| **Report Date** | | 1 May 2017 |
| **Chief Investigator / Supervisor** | | Dr Helen Healy |
| **Administering Institution** | | The University of Queensland |
| **Funding Period** | Start Date: | 1 December 2015 |
| | Finish Date: | 1 December 2016 |

## PROJECT SUMMARY

This PhD program of work seeks to translate advances in genetic technologies into healthcare for those affected by inherited kidney disease.

Firstly, this involves undertaking epidemiological analyses of cohorts of patients in Australia affected by inherited kidney diseases individually and collectively.

Secondly, a clinical and multidisciplinary service model has been proposed and commenced with short-term evaluation of effectiveness to be undertaken.

Thirdly, a collaborative diagnostic genomic service is being established, validated and assessed to identify causative variants in known genes implicated in inherited kidney disease for affected Australian families.

Lastly, a research genomic and functional genomic project has been assessed in order to identify and validate new or novel genetic causes of inherited kidney disease for those in whom current diagnostic genomics has been unable to secure a genetic diagnosis.

Together this pipeline of activities is a patient-centric attempt to understand and explain inherited kidney disease in Australia whilst investigating ways in which to optimise clinical care and to thus attempt to improve future outcomes.

## PROJECT AIMS / OBJECTIVES

The hypothesis of this study is that modern genetic sequencing and interpretation in GRD can contribute to personalised care by

Aim 1 Systematic Reviews of

- Clarifying clinical diagnosis
- Improving options for clinical management

Aim 1.1 Therapeutics in GRD

Aim 1.2 GRD Clinical Services

Aim 2 Profiling GRD populations

Aim 2.1 Registry-based analysis of existing GRD cohorts

Aim 2.2 Identify cohort with Phenotypically Indeterminate GRD

Aim 2.3 Describe phenotypes/pedigrees

Aim 3 Genetic Sequencing of GRD populations

Aim 3.1 DNA extraction/banking

Aim 3.2 Perform Whole Exome Sequencing (WES)

Aim 3.3 Describe and interpret gene mutations of affected patients and families

Aim 4 Translation into Personalised Healthcare

Aim 4.1 Validate GRD genotype-phenotype relationships against clinical outcomes

Aim 4.2 Implement Collaborative GRD Service

Aim 4.3 Identify candidates for targeted therapeutics

## SIGNIFICANCE AND OUTCOMES

This PhD program has this year resulted in several significant outcomes:

1. Commencement and growth of RBWH Renal Genetics Clinic (commenced 1 /7 /13) - a national first

2. First Description of the Epidemiology of Genetic Renal Disease in QLD - publication OJRD

3. Report of the Epidemiology of RRT treated Al port Syndrome related ESKD in Australia and New Zealand – publication NDT

4. Dissemination of a proposed Australian Renal Genetic Clinic Model - abstracts ANZSN, HGSA, RBWH Symposium

5. Commencement of Diagnostic Renal Gene Panels - abstracts ANZSN, ASN, RBWH Symposium (Award – Best Clinical Science Oral)

6. Commencement of a national Research & Functional Genomics Project for cases of genetically unresolved inherited kidney disease

## PUBLICATIONS / PRESENTATIONS

Publications during candidature

Published Peer-Reviewed Articles

1. Francis A, Burke J, Francis L, McTaggart S, Mallett A. Polypoid change of the glomerular basement membrance in a child with steroid resistant nephrotic syndrome and ARHGAP24 mutation: a case report. Open Journal of Uro logy and Nephrology, accepted Dec 2015, In Press, doi: pending.

2. Mallett A, Fowles LF, McGaughran J, Healy H, Patel C. A Multidisciplinary Renal Genetics Clinic improves patient diagnosis. Medical Journal of Australia, Feb 2016; 204(2):58-59.
3. Rangan G, Alexander SI, Campbell K, Dexter M, Lee V, Lopez- Vargas P, Mai J, Mallett A, Patel C, Patel M, Tunnicliffe D, Tehan M, Tong A, Vladica P, Savige J. KHA-CARI guideline recommendations for the diagnosis and management of autosomal dominant polycystic kidney disease. Nephrology, accepted Oct 2015, In Press, doi: 10.1111 /nep.12658.
4. Mallett A, Lee V, Mai J, Lopez-Vargas P, Rangan G. KHA-CARI Autosomal-Dominant Polycystic Kidney Disease Guideline: Pharmacological Management. Seminars in Nephrology, Nov 2015; 35(6):582-589.
5. Mallett A, Patel M, Tunnicliffe D, Rangan G. KHA-CARI Autosomal-Dominant Polycystic Kidney Disease Guideline: Management of Renal Stone Disease. Seminars in Nephrology, Nov 2015; 35(6):603-606.
6. Patel C, Tehan M, Savig J, Mallett A, Tong A, Tunnicliffe D, Rangan G. KHA-CARI Autosomal-Dominant Polycystic Kidney Disease Guideline: Genetics and Genetic Counselling. Seminars in Nephrology, Nov 2015; 35(6):550-556.
7. Tehan M, Savige J, Patel C, Mallett A, Tong A, Tunnicliffe D, Tangan G. KHA-CARI Autosomal-Dominant Polycystic Kidney Disease Guideline: Genetic Testing in Diagnosis. Seminars in Nephrology, Nov 2015; 35(6):545-549.
8. Savige J, Mallett A, Tunnicliffe D, Rangan G. KHA-CARI Autosomal-Dominant Polycystic Kidney Disease Guideline: Management of Polycystic Liver Disease. Seminars in Nephrology, Nov 2015; 35(6):618-622.
9. Tong A, Mallett A. Lopez-Vargas P, Rangan G. KHA-CARI Autoso1 Dominant Polycystic Kidney Disease Guideline: Psychosocial Care. Seminars in Nephrology, Nov 2015; 35(6):590-594.
10. Mallett A, Patel C, Maier B, McGaughran J, Gabbett M, Takasato M, Cameron A, Trnka P, Alexander SI, Rangan G, Tehan M, Caruana G, Quinlan C, McCarthy H, Hyland V, Hoy W, Wolvetang E, Taft R, Simons C, Healy H, Little M. A protocol for the identification and validation of novel genetic causes of kidney disease. BMC Nephrology, Sept 2015; 16(1):152.
11. Mallett A, Hughes P, Szer J, Tuckfield A, Van Eps C, Campbell SB, Hawley C, Burke J, Kausman J, Hewitt I, Parnham A, Ford S, Isbel N. Atypical Haemolytic Uraemic Syndrome treated with the complement inhibitor Eculizumab: the experience of the Australian Compassionate access cohort. Internal Medicine Journal, Nov 20 15; 45(10):1054-65.
12. Mallett A, Corney C, McCarthy H, Alexander SI, Healy H. Genomics in the Renal Clinic - Translating nephrogenetics for clinical practice. Human Genomics, June 2015; 9(1 3).
13. Mallett A, Tang W, Hart G, McDonal S, Hawley C, Badve S, Boudville N, Brown F, Campbell S, Clayton P, Johnson D. Endstage kidney disease due to fibrillary glomerulonephritis and immunotactoid glomerulopathy- Outcomes in 66 consecutive ANZDATA Registry cases. American Journal of Nephrology, Sept 20 15; 32(3):177-84.
14. Tong A, Tunnicliffe D, Lopez-Vargas P, Mallett A, Patel C, Savige J, Campbell K, Patel M, Tehan M, Alexander SI, Lee V, Craig JC, Fassett R, Rangan G. Identifying and integrating consumer perspectives in clinical practice guidelines on autosomal dominant polycystic kidney disease. Nephrology, accepted Jul 2015, In Press, doi: 10.1111/nep.1 2579.
15. Ying T, Hill P, Desmond M, Agar J, Mallett A. Fibrillary Glomerulonephritis: an apparent familial form? Nephrology, July 2015; 20(7):506-9.

16. Mallett A, Sandford R. NOS3 as a Potential Modifier of ADPKD Phenotypic Variability - Progress towards an answer. Nephrology journal, Dec 2014; 19(12):733-4.
17. Mallett A, Tang W, Clayton P, Stevenson S, McDonald S, Hawley C, Badve S, Boudville N, Brown F, Campbell SB, Johnson D. End-stage kidney disease due to Alport Syndrome - Outcomes in 296 consecutive ANZDATA Registry cases. Nephrology Dialysis Transplantation, July 2014; 29(12):2277-86.
18. Mallett A, Patel C, Salisbury A, Wang Z, Healy H, Hoy W. The prevalence and epidemiology of genetic renal disease amongst adults with chronic kidney disease in Australia. Orphanet Journal of Rare Disease, June 2014; 9(98).
19. Stevenson S, Mallett A, Oliver K, Hyland V, Hawley C, Malmanche T, Isbel, N. Atypical HUS associated with severe, unexpected antibody mediated rejection post kidney transplant. Nephrology, April 2014; 19(Supp1):22-6.

Manuscripts Under Review

1. Connor T, Hoer S, Mallett A, Gale D, Gomez-Duran A, Posse V, Antrobus P, Moreno P, Sciavelli M, Frezza C, Duff J, Sheerin N, Sayer J, Ashcroft M, Wiesener M, Gustafsson C, Hudson G, Chinnery P, Maxwell P. A homoplasmic mitochochondrial DNA control region mutation causes tubulointerstitial kidney disease. Nature Genetics, submitted August 2016.
2. Mallett A, McCarthy H, Ho G, Holman K, Farnsworth E, Patel C, Fletcher J,, Mallawaarachchi A, Quinlan C, Bennetts B, Alexander SI. A targeted exomic approach achieves genetic diagnosis for multiple nephrogenetic phenotypes in a diagnostic genetic renal disease sequencing service. Kidney International, pending submission October 2016.

Conference Abstracts - Oral Presentations

1. Mallett A (oral presenter), Maier B, Er P, Takasato, M, Sun J, Wolvetang E, Alexander S, Simons C, Little M. Patient-Derived Induced Pluripotent Stem Cell (iPSC) Modeling of Genetic Renal Disease (GRD).

    a. 2015 American Society of Nephrology Kidney Week, San Diego (USA), November 2015. Published as JASN; 26(S1):1 2a.

    b. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium.

2. Mallett A (oral presenter), Patel C, Crawford J, Bennetts B, Little M, Healy H, Alexander S, Hyland V, Simons C. Massively Parallel Sequencing (MPS) in Diagnostically Refractory Genetic Renal Disease (GRD).

    a. 2015 American Society of Nephrology Kidney Week, San Diego (USA), November 2015. Published as JASN; 26(S1):46a.

3. Little M (oral presenter), Wolvetang E, Maier B, Sun J, Er P, Takasato M, Mallett A, Alexander S, Bennetts B. Modelling genetic kidney disease using patient-derived and CRISPR-Cas9 generated mutant pluripotent stem cells.

    a. 2015 American Society of Nephrology Kidney Week - Early Program: Advances in Research Conference "Engineering Genomes to Model Disease, Target Mutations and Personalise Therapy", San Diego (USA), November 2015.

4. Mallett A (oral presenter), Mordaunt D, Crafter S, McTaggart S, Kark A, Patel C, Crawford J, Holman K, Farnsworth E, Ho G, Healy H, Alexander SI, Bennetts B, Little M, Simons C. The heterozygous p.R76W HNF4A variant is associated with atypical autosomal dominant de Toni-Fanconi-Debre Syndrome and can be diagnosed utilizing diagnostic clinical exomic analysis.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):41.

5. Mallett A (oral presenter), Mordaunt D, Sonawane R, Walker A, Kausman J, Peters H, White S, Stark Z, Trnka P, Patel C, Crawford J, Holman K, Farnsworth E, Ho G, Alexander S, Bennetts B, Healy H, Little M, Simons C, Yaplito-Lee J. RMND1 mutations are associated with autosomal recessive syndromic nephropathy.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):42.

6. Mallett A (oral presenter), Hoer S, John G, Burke J, Patel C, Crawford J, Hyland V, Healy H, Little M, Simons C, Connor T, Maxwell P. The T616C tRNA(Phe) mutation causes mitochondrially inherited tubulointerstitial kidney disease.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):41.

b. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium, October 20 15, Prize: "2015 RBWH Foundation Award for Best Overall Research"

7. Tunnicliffe D (oral presenter), Tong A, Lopez-Vargas, Mallett A, Patel C, Savige J, Campbell K, Patel M, Tehan M, Alexander S, Lee V, Craig J, Rangan G. Identifying and integrating consumer perspectives in clinical practice guidelines on autosomal dominant polycystic kidney disease.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):51.

8. Little M (oral presenter), Takasato M, Er P, Chiu H, Maier B, Wolvetang E, Sun J, Simons C, Mallett A. The directed differentiation from pluripotent cells to kidney cells.

a. 2015 International Workshop on Development Nephrology, Snowbird (USA), July 201 5.

9. Mallett A (oral presenter), Campbell S, Van Eps C, Hawley C, Burke J, Hughes P, Kausman J, Hewitt I, Parnham A, Szer J, Tuckfield A, Payne S, Young J, Isbel N. Eculizumab is effective therapy for atypical haemolytic uraemic syndrome (aHUS): a case series of Australian patients.

a. 2014 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S1):37.

10. Mallett A (oral presenter), Ho G, McCarthy H, Fletcher J, Mallawaarachchi A, Little M, Juepner H, Sawyer A, Bennetts B, Alexander S. Exomic approaches to diagnosis amongst Australians with Genetic Renal Diseases.

a. 2014 Royal Brisbane and Women's Hospital Symposium, October 2014, Prize: "Best Clinical Science Oral Presentation"

11. Patel C (oral presenter), Mallett A, Healy H, McCaughran J. The Queensland renal genetics multidisciplinary team clinical service.

a. 2014 Human Genetics Society of Australasia, Adelaide, August 2014.

12. Mallett A (oral presenter), Salisbury A, Wang Z, Healy H, Hoy W. Autosomal Dominant Polycystic Kidney Disease in an Australian Chronic Kidney Disease (CKD) Population.

a. 2013 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Brisbane, September 2013. Published as Nephrology; 18(S1)47.

13. Mallett A (oral presenter), Salisbury A, Wang Z, Healy H, Hoy W. Alpot1 Syndrome and Thin Basement Membrane Nephropathy in the Queensland Chronic Kidney Disease (CKD) Registry.

a. 2013 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Brisbane, September 2013. Published as Nephrology; 18(S1 )23.

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 201 3.

14. Mallett A (oral presenter), Salisbury A, Wang Z, Healy H, Hoy W. Acute Kidney Injury, Analgesic Nephropathy and ToxinMediated Kidney Injury in an Australian Chronic Kidney Disease (CKD) Cohot1.

a. 2013 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Brisbane, September 2013. Published as Nephrology; 18(S 1 )42.

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

15. Healy H (oral presenter), Salisbury A, Wang Z, Mallett A, Huynh S, Salisbury A, Mohandas T, Sanghi P, Heffernan D, Fassett R, Hoy W. Chronic Kidney Disease (CKD) is not Renal Replacement Therapy (RRT) - The CKD. QLD Registry dataset.

a. 2013 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Brisbane, September 2013. Published as Nephrology; 18(S1 )33.

16. Salizbury A (oral presenter), Mallett A, Wang Z, Healy H, Huynh S, Smith S, Heffernan D, Hoy W. Chronic Kidney Disease (CKD) Patient Outcomes: A longitudinal report from the CIW. QLD registry.

a. 2013 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Brisbane, September 2013. Published as Nephrology; 18(S 1 )32.

Conference Abstracts - Poster Presentations

1. Mallett A, Healy H, McGaughran J, Little M, Patel C. Characteristics and Initial Outcomes of a Multidisciplinary Renal Genetics Clinic (RGC)

a. 2015 American Society of Nephrology Kidney Week, San Diego (USA), November 2015. Published as JASN; 26(S1 ):995A.

2. Wilson G, Kark A, Mallett A, Salisbury A, Wang Z, Healy H, Hoy W. Effects of Acute Kidney Injury (AKI) Severity and CoMorbidities on Chronic Kidney Disease (CKD) Progression

a. 2015 American Society of Nephrology Kidney Week, San Diego (USA), November 2015. Published as JASN; 26(S1 ):480A.

3. Wilson G, Kark A, Mallett A, Cameron A, Wang Z, Kirby J, Healy H, Hoy W. Acute Kidney Injury (AKI) associated with Chronic Kidney Disease (CKD) in the renal practices of the Royal Brisbane and Women's Hospital (RBWH) through the CKD.QLD registry.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):74.

b. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium, October 2015.

4. Mallett A, Patel C, Crawford J, Bennetts B, Little M, Healy H, Alexander S, Hyland V, Simons C. Massively Parallel Sequencing (MPS) in Diagnostically Refractory Genetic Renal Disease (GRD).

a. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium, October 2015.

5. Mallett A, Mordaunt D, Crafter S, McTaggart S, Kark A, Patel C, Crawford J, Holman K, Farnsworth E, Ho G, Healy H, Alexander SI, Bennetts B, Little M, Simons C. The heterozygous p.R76W HNF4A variant is associated with atypical autosomal dominant de Toni-Fanconi-Debre Syndrome and can be diagnosed utilizing diagnostic clinical exomic analysis.

a. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium, October 2015.

6. Mallett A, Mordaunt D, Sonawane R, Walker A, Kausman J, Peters H, White S, Stark Z, Trnka P, Patel C, Crawford J, Holman K, Farnsworth E, Ho G, Alexander S, Bennetts B, Healy H, Little M, Simons C, Yaplito-Lee J. RMND1 mutations are associated with autosomal recessive syndromic nephropathy.

a. 2015 Royal Brisbane and Women's Hospital Healthcare Symposium, October 2015.

7. Mallawaarachchi A, Fang H, Sadowski C, Mallett A, McCarthy H, Clayton P, Rangan G, O'Connell P, Lewis D, Ho G, Bennetts 8, Hildebrandt F, Alexander S, Tehan M. X-Jinked CLN5 variants in a multi-generational family with variable age-of-onset of renal failure and proteinuria.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):79.

8. Mahmood U, Hoy W, Kark A, Healy H, Mallett A, Rawlings C, Wang Z, Kirby J, Cameron A. Heterogeneity of Chronic Kidney Disease (CKD) by age in an Australian Metropolitan Renal Service.

a. 201 5 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):65.

9. Mahmood U, Hoy W, Kark A, Healy H, Mallett A, Rawlings C, Wang Z, Kirby J, Coleman S, Cameron A. Profiles of very elderly patients with Chronic Kidney Disease(CKD) in the public renal specialty practices of the Royal Brisbane and Women's Hospital (RBWH) in Queensland.

a. 2015 Australian and New Zealand Society of Nephrology Annual Scientific Meeting, Canberra, September 2015. Published as Nephrology; 20(S3):65.

10. Maier B, Er P, Takasato M, Wolvetang E, Simons C, Mallett A, Little M. Patient iPSC-based modelling of genetic renal disease.

a. 2015 Australasian Society for Stem Cell Research, Hunter Valley, November 2015.

b. 2015 International Society for Stem Cell Research, Stockholm, June 2015.

11 . Mordaunt D, Crafter S, Coates D, Haan E, McTaggart S, Karka A, Crawford C, Holman K, Simons C, Alexander S, Ho G, Bennetts B, Lillie M, Mallett A. Patients with atypical dominant De ToniFanconi- Debre syndrome, perturbations in glucose metabolism and mu/tisystem involvement associated with the heterozygous R76W variant in HNF4A.

a. 2015 Human Genetics Society of Australasia Annual Scientific Meeting, Perth, August 2015.

12. Mallett A, Campbell S, Van Eps C, Hawley C, Burke J, Hughes P, Kausman J, Hewitt I, Parnham A, Szer J, Tuckfield A, Payne S, Young J, Isbel N. Eculizumab is effective therapy for atypical haemolytic uraemic syndrome (aHUS): a case series of Australian patients.

a. 2014 Royal Brisbane and Women's Hospital Healthcare Symposium, October 2014.

13. Mallett A, Patel C, McGaughran J, Healy H. The initial six months of an Australian Renal Genetics Clinic Service.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourn e, August 2014. Published as Nephrology; 19(S4 ):68.

14. Mallawaarachchi A, Mallett A, Sawyer A, McCarthy H, Fletcher J, Chapman J, Bennetts 8, Ho G, Juepner H, Hahn D, Alexander S. Utilising Exome Sequencing to identify nephronophthisis mutations within an Australian clinical cohort.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4):71.

15. Fletcher J, Mallett A, Ho G, McCarthy H, Sawyer A, Mallawaarachchi A, Rosier M, Little M, Bennetts B, Juepner H, Turner A, Alexander S. Heterozygous LMX1B mutation detection in familial FSGS without extrarenal manifestations using whole exome seque a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4 ):69.

16. McCarthy H, Sawyer A, Fletcher J, Mallett A, Mallawaarachchi A, Ho G, Bennetts B, Juepner H, Alexander S. Whole exome sequencing identifies a novel mutation in A TP6VOA4 in familial distal renal tubular acidosis.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4 ):67.

17. Mallett A, Ho G, McCarthy H, Fletcher J, Mallawaarachchi A, Little M, Juepner H, Sawyer A, Bennetts B, Alexander S. Exomic approaches to diagnosis amongst Australians with Genetic Renal Diseases.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4 ):68.

18. Rawlings C, Francis L, Mallett A, John G, Denaro C. Coincident lgA Nephropathy in an Australian patient with Fabry's Disease.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4 ):94.

19. Rawlings C, Susman R, Mallett A, Francis L, Kark A. Renal Oncocytosis in the setting of a rare unvalidated FLCN gene variant.

a. 2014 Australian and New Zealand Annual Scientific Meeting, Melbourne, August 2014. Published as Nephrology; 19(S4 ):94.

20. Mallett A, Campbell S, Van Eps C, Hawley C, Burke J, Hughes P, Kausman J, Hewitt I, Parnham A, Szer J, Tuckfield A, Payne S, Young J, Isbel N. Eculizumab is effective therapy for atypical haemolytic uraemic syndrome (aHUS): a case series of Australian patients.

a. 2014 Annual Scientific Meeting of the Haematology Society of Australia and New Zealand, the Australian & New Zealand Society of Blood Transfusion and the Australasian Society of Thrombosis and Haemostasis, Perth, October 2014.

21 . Mallett A, Ho G, McCarthy H, Little M, Benentts B, Alexander S. Clinical diagnostic testing amongst Australians with Genetic Renal Diseases using a Targetted Exomic Approach.

a. 2014 American Society of Nephrology Kidney Week, Philadelphia (USA), November 2014. Published as JASN; 25(S1 ):422A.

22. Healy H, Wang Z, Mallett A, Huynh S, Coleman S, Kark A, Salisbury A, Venuthurupalli S, Fassett R, Hoy W. Chronic Kidney Disease (CKD) is a different population - The CKD.QLD Registry

dataset.

a. 2013 Royal Australasian College of Physicians Annual Scientific Meeting, Perth, May 2013. Published as Internal Medicine Journal; 43(S3):42.

23. Healy H, Mallett A, Wang Z, Salisbury A, Fassett R, Hoy W. Chronic Kidney Disease (CKD) is distinct from End Stage Kidney Disease (ESKD).

a. 2013 American Society of Nephrology Kidney Week, Atlanta (USA), November 2013. Published as JASN; 24(1 ):893A.

b. 2013 Royal Brisbane and Women's Hospital Symposium,ncing. October 2013.

17. Mallett A, Salisbury A, Wang Z, Healy H, Hoy W. A/port Syndrome and Thin Basement Membrane Nephropathy in the Queensland Chronic Kidney Disease (CKD) Registry.

a. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

18. Mallett A, Salisbury A, Wang Z, Healy H, Hoy W. Acute Kidney Injury, Analgesic Nephropathy and Toxin-Mediated Kidney Injury in an Australian Chronic Kidney Disease (CKD) Cohort.

a. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

24. Mallett A, Salisbury A, Wang Z, Healy H, John G, Hoy W. Autosomal Dominant Polycystic Kidney Disease (ADPKD) and Medullary Cystic Kidney Disease (MCKD) in an Australian Chronic Kidney Disease (CKD) Cohort.

a. 2013 American Society of Nephrology Kidney Week, Atlanta (USA), November 2013. Published as JASN; 24(1 ):304A.

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

25. Mallett A, Salisbury A, Wang Z, Healy H, John G, Hoy W. Characteristics of Uninephric Chronic Kidney Disease (CKD) patients.

a. 2013 American Society of Nephrology Kidney Week, Atlanta (USA), November 2013. Published as JASN; 24(1 ):685A.

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

26. Mallett A, Salisbury A, Wang Z, Healy H, John G, Hoy W. Glomerulonephritis (GN) in a Chronic Kidney Disease (CKD) Population.

a. 2013 American Society of Nephrology Kidney Week, Atlanta (USA), November 2013. Published as JASN; 24(1):824A

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

27. Healy H, Mallett A, Salirbury A, Wang Z, Hoy W. Clinical Outcomes in Australian Chronic Kidney Disease (CKD) Patients. a. 2013 American Society of Nephrology Kidney Week, Atlanta (USA), November 2013. Published as JASN; 24(1):677A.

b. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.

28. Rawlings C, Mallett A, Wang Z, Healy H, Salisbury A, Hoy W. Number of admissions of patients with chronic kidney disease predicts risk of death and renal replacement therapy.

a. 2013 Royal Brisbane and Women's Hospital Symposium, October 2013.