

Case report: Central 22q11.2 Deletion Presenting in Adulthood with CAKUT, Malignant Hypertension and Thoracoabdominal Aortopathy

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Case Report

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Abstract

Background

Congenital anomalies of the kidney and urinary tract (CAKUT) are among the leading causes of chronic kidney disease (CKD) and are increasingly recognized to have a genetic basis. Pathogenic copy number variants (CNVs), particularly those involving chromosome 22q11.2, represent important but frequently underrecognized causes of syndromic CAKUT. Central 22q11.2 deletions, which spare *TBX1* while encompassing *CRKL*, often lack the classic manifestations of DiGeorge syndrome and may therefore remain undiagnosed until adulthood.

Case presentation

We report a 40-year-old woman with congenital solitary functioning kidney secondary to left kidney hypoplasia, longstanding resistant hypertension, and ischemic stroke in the setting of malignant hypertension. Vascular imaging demonstrated multifocal narrowing of the descending thoracic and abdominal aorta with extensive collateralization, consistent with developmental arteriopathy. Additional findings included craniofacial and dental abnormalities, learning difficulties, and a family history of chronic kidney disease, stroke and developmental delay. Comprehensive genomic testing using a CAKUT-focused next-generation sequencing panel with integrated CNV analysis identified a heterozygous pathogenic 954.8-kb central 22q11.21 deletion spanning the LCR22B–D interval and encompassing *CRKL* while sparing *TBX1*.

Conclusions

This case expands the adult phenotype associated with central 22q11.2 deletion syndrome and illustrates how developmental kidney and vascular abnormalities may remain unrecognized for decades in the absence of the classic features of DiGeorge syndrome. Recognition of the underlying genomic disorder reclassified the patient's vascular disease as a developmental arteriopathy rather than an acquired inflammatory process, with important implications for surveillance, therapeutic decision-making, genetic counseling, and cascade testing. Central 22q11.2 deletions are an underrecognized cause of syndromic CAKUT in adults and as illustrated, may also underlie developmental large-vessel arteriopathy. Adults presenting with CAKUT, severe or resistant hypertension, unexplained large-vessel abnormalities, or additional extra-renal manifestations should undergo comprehensive genomic evaluation that includes CNV analysis to establish a molecular diagnosis and guide clinical management.

Background

Congenital anomalies of the kidney and urinary tract (CAKUT) comprise a heterogeneous group of developmental disorders affecting the kidneys and urinary tract and represent one of the leading causes of chronic kidney disease (CKD) (1, 2). Advances in genomic medicine have demonstrated that approximately 10–20% of individuals with CAKUT harbor an identifiable monogenic disorder or pathogenic copy number variant (CNV), with substantially higher diagnostic yields among patients with syndromic or extra-renal manifestations (2, 3). Despite these advances, many individuals with mild congenital kidney anomalies survive into adulthood without an etiologic diagnosis, particularly when syndromic features are subtle (4, 5).

Recurrent CNVs involving chromosome 22q11.2 represent an important cause of syndromic CAKUT and illustrate the critical role of dosage-sensitive developmental genes in human nephrogenesis (6, 7). The genomic architecture of the 22q11.2 locus, characterized by multiple low-copy repeats (LCR22A–H), predisposes to recurrent rearrangements through non-allelic homologous recombination (8). Proximal deletions involving *TBX1* cause classic 22q11.2 deletion syndrome (DiGeorge syndrome), which is typically recognized during infancy or childhood because of congenital heart disease, thymic and parathyroid hypoplasia, palatal abnormalities, immunodeficiency, and characteristic craniofacial features (9).

By contrast, central 22q11.2 deletions, which spare *TBX1*, constitute a distinct genomic disorder with a predominance of renal and urinary tract malformations, developmental delay and cognitive or behavioural difficulties, with congenital heart disease seen less frequently than in the common proximal deletion (10, 11). Increasing evidence identifies haploinsufficiency of *CRKL* as the principal developmental driver of the renal phenotype, disrupting signaling pathways essential for ureteric bud branching and nephron formation (6, 7). Consequently, affected individuals may present primarily with congenital kidney anomalies, hypertension, or vascular disease, often without the classic manifestations of DiGeorge syndrome, delaying diagnosis until adulthood (10, 12).

Here we report an adult who presented with congenital solitary functioning kidney, malignant hypertension, ischemic stroke, and thoracoabdominal developmental arteriopathy, in whom comprehensive genomic testing identified a pathogenic central 22q11.21 deletion involving *CRKL*. This case illustrates the importance of incorporating CNV analysis into nephro-genetic evaluation of adults with CAKUT and demonstrates how establishing a molecular diagnosis can alter diagnostic interpretation, clinical management, and family counseling (6, 13).

Case presentation

A 40-year-old woman with a history of congenital left kidney hypoplasia, treated with nephrectomy at 5 months of age, was referred for nephro-genetic assessment following an ischemic cerebrovascular event in the setting of malignant hypertension. She was born two weeks post-term with a birth weight of 2.1 kg. Early growth and development were otherwise unremarkable. A history of learning difficulties requiring educational support was noted although to our knowledge no formal assessment or diagnosis

was ever performed. Family history was notable for her father who had a stroke and died from kidney failure and a brother with developmental delay.

Hypertension was diagnosed at age 26 and remained suboptimally controlled despite multidrug therapy. Kidney function was stable for many years (serum creatinine 120–140 $\mu\text{mol/L}$) with minimal proteinuria (urine albumin creatinine ratio of 0.6 mg/mmol). Physical examination identified sparse eyebrows and a short neck. She also reported lifelong dental abnormalities with premature tooth loss.

At 39 years, she presented with acute right-sided weakness and dysarthria. Blood pressure was markedly elevated (up to 251/188 mmHg). Neuroimaging demonstrated an acute lacunar infarct in the left corona radiata without large-vessel occlusion. Stroke etiology was attributed to small-vessel disease in the setting of severe hypertension. Cardiac evaluation revealed concentric left ventricular hypertrophy with preserved systolic function and no arrhythmia or shunt. Imaging showed no evidence of carotid or vertebral stenosis. Functional outcome at discharge corresponded to a modified Rankin Scale score of 2.

During hospitalization, hypertension remained resistant despite therapy escalation. Laboratory investigations showed stable CKD (serum creatinine 155 $\mu\text{mol/L}$), normal electrolytes, and minimal albuminuria. Evaluation for secondary hypertension, including endocrine and renovascular causes, was unremarkable. Inflammatory markers were normal. CT angiography demonstrated multifocal narrowing of the descending thoracic and upper abdominal aorta, with minimal luminal diameters of 5–7 mm, post-stenotic dilatation, and extensive collateralization via the pathway of Winslow with dilation of the internal thoracic and inferior epigastric arteries (Fig. 1). Renal arteries were normal. The patient reported claudication occurring after approximately one kilometer. Femoral, popliteal, and pedal pulses were absent. Ankle–brachial indices were reduced (right 0.52, left 0.51), consistent with severe proximal inflow disease. Physical examination demonstrated ischemic rest pain without tissue loss. Capillary refill was adequate. Arterial Doppler examination demonstrated monophasic waveforms throughout the lower extremities.

The differential diagnosis included large-vessel vasculitis, fibromuscular dysplasia, congenital coarctation variants, and genetic arteriopathy within a mid-aortic syndrome spectrum. Acquired inflammatory etiologies were considered less likely given normal inflammatory markers and extensive collateralization.

Given the combination of CAKUT, severe hypertension, vascular abnormalities and syndromic features with a positive family history of CKD, genomic testing was pursued (Fig. 2). Genomic DNA was extracted from whole blood (EDTA). Targeted next-generation sequencing of a CAKUT-focused gene panel, including the coding regions of the targeted genes and ± 10 bp of intronic flanking sequence, was performed using hybridization capture followed by sequencing on the Illumina NovaSeq 6000 platform (2×150 bp; Prevention Genetics) (14) (**Table S1**). Quality metrics included $> 98\%$ of target bases covered at $\geq 20\times$ and a mean coverage depth $> 100\times$. Sequence data were analyzed using Prevention Genetics' internally developed bioinformatic pipeline, with Sanger sequencing used to supplement regions with

insufficient coverage when applicable (14, 15). Copy number variants were inferred from NGS read-depth data, with analytical sensitivity approaching 100% for events spanning four or more exons (14, 15).

Testing identified a heterozygous pathogenic deletion of approximately 954.8 kb at chromosome 22q11.21, with a minimum boundary defined by chr22:20,459,997–21,414,845 (GRCh37/hg19) (**Table S2**). The deletion corresponds to the recurrent low-copy repeat–mediated interval spanning LCR22B–D and spares the *TBX1* gene locus (Fig. 2) (9, 10). The CNV is classified as pathogenic according to American College of Medical Genetics and Genomics criteria (16). The deleted interval encompasses 15 protein coding genes (**Table S2**). Parental DNA was not available; therefore, inheritance could not be determined. Given the family history of kidney failure and cerebrovascular disease in her father, cascade testing of first-degree relatives was recommended.

Discussion and Conclusions

This case describes an adult presentation of a central 22q11.2 deletion involving *CRKL*, characterized by a kidney- and vascular-dominant phenotype. In contrast to proximal 22q11.2 deletion syndrome, which is typically identified in childhood because of cardiac, immunologic, and endocrine manifestations (9), central deletions frequently lack these classic features and may remain unrecognized until adulthood (10). The patient's additional features, sparse eyebrows, a short neck, dental abnormalities, and learning difficulties, may be consistent with the variable extra-renal manifestations described in central 22q11.2 deletion, supporting a single unifying genomic diagnosis rather than coincidental comorbidity.

Haploinsufficiency of *CRKL* is considered the principal driver of the kidney and vascular phenotype. *CRKL* encodes an adaptor protein mediating intracellular signaling downstream of receptor tyrosine kinases, including fibroblast growth factor receptors (FGFRs). It plays a critical role in ureteric bud branching morphogenesis, nephron progenitor differentiation, pharyngeal arch development, and neural crest derived vascular formation (6, 7, 17). Experimental models demonstrate that *Crkl* deficiency leads to renal hypodysplasia and ureteral abnormalities through impaired signaling at the metanephric mesenchyme-ureteric bud interface, as well as aortic arch defects arising from neural crest dysfunction (6, 18, 19). Therefore, disruption of these pathways provides a mechanistic basis for the combination of congenital kidney hypoplasia and large-vessel abnormalities observed in this patient.

The vascular phenotype, which includes multifocal narrowing of the descending thoracic and upper abdominal aorta with collateralization and normal inflammatory markers, is most consistent with a developmental arteriopathy rather than acquired large-vessel vasculitis. To our knowledge, thoracoabdominal aortopathy of this type has not previously been reported in a central 22q11.2 deletion. A developmental basis is plausible: in cardiac neural crest cells, *CRKL* together with *CRK* contributes to vascular smooth muscle differentiation, and *Crkl* loss causes aortic arch artery defects in mice (18, 19). However, cardiac neural crest forms the ascending aorta and arch, whereas the descending thoracic aorta is derived from the paraxial mesoderm and the abdominal aorta is derived from non-neural-crest

mesoderm (20); the distribution here therefore suggests CRKL may influence vascular development beyond the neural crest derived segments, however this finding warrants further evaluation.

This case highlights several important clinical implications. First, congenital kidney hypoplasia treated with nephrectomy in childhood should be recognized within the CAKUT spectrum rather than as an isolated historic finding. Individuals with solitary kidneys due to congenital anomalies are at increased risk of hypertension and kidney dysfunction (21, 22). Second, coexistence of CAKUT and large-vessel abnormalities, as an extra-renal manifestation of disease, warrants consideration of an underlying genetic etiology (1). Third, sequence-based testing alone is insufficient in this context and CNV analysis is essential to detect pathogenic dosage alterations especially when there is a combined renal and vascular phenotype (4, 9).

Importantly, establishing a molecular diagnosis reframed this patient's vascular disease as a developmental genetic process rather than a primary inflammatory vasculitis. This distinction has direct therapeutic implications, avoiding unnecessary immunosuppressive therapy and redirecting management toward vascular surveillance and aggressive blood pressure control.

Finally, central 22q11.2 deletions follow an autosomal dominant inheritance pattern with variable expressivity. Identification of a pathogenic CNV enables appropriate genetic counseling and cascade testing, particularly in the presence of a family history suggestive of heritable disease.

Although limited by descriptive nature of a single case report and the inability to perform familial segregation analysis, this report highlights an underrecognized presentation that may facilitate earlier genomic diagnosis in similar patients. Segregation analysis would have been of particular interest given the family history of kidney failure, stroke and developmental delay, and given that central 22q11.2 deletions are more frequently inherited compared to a predominant de novo inheritance pattern in DiGeorge Syndrome (10). In addition, the vascular phenotype described here is a single observation and requires confirmation in further cases before a causal link to CRKL haploinsufficiency can be confirmed.

Central 22q11.2 deletions involving *CRKL* are an underrecognized cause of combined renal and vascular disease presenting in adulthood. Adults with CAKUT, refractory or malignant hypertension and unexplained large-vessel abnormalities may harbor pathogenic CNVs despite absence of classic syndromic features. Molecular diagnosis informs management, avoids inappropriate immunosuppression, and enables genetic counseling. Incorporation of CNV analysis into nephro-genetic evaluation is essential to establish diagnosis, guide management, and enable family screening.

Abbreviations

CAKUT

Congenital anomalies of the kidney and urinary tract

CKD

Chronic kidney disease

CNV
Copy number variant
CRKL
CRK-like proto-oncogene, adaptor protein
CT
Computed Tomography
FGFR
Fibroblast growth factor receptor
LCR
Low-copy repeat
MIP
maximum intensity projection
NGS
Next-generation sequencing

Declarations

Ethics approval and consent to participate

The procedures followed in this study were approved by the Western University Research Ethics Board (REB #115160) and were conducted in accordance with the Declaration of Helsinki. Written informed consent was obtained from the participant for participation in this study.

Consent for publication

Written informed consent was obtained from the participant for publication of this case report, including the clinical details and accompanying images.

Availability of data and materials

The genomic findings reported were generated as part of routine clinical diagnostic testing rather than research testing. The targeted panel sequencing data, including BAM and VCF files, are not released to us and therefore cannot be deposited in a controlled-access repository. In addition, the participant's consent and institutional ethics prohibits the release of such data. In the context of a single case report, such data could contain potentially identifiable patient information. Additional de-identified information relevant to this case may be made available from the corresponding author upon reasonable request, subject to institutional approvals and data transfer agreements, where applicable.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

MA and DMC conceived the study. MA collected and interpreted the clinical data, performed the literature review, and drafted the manuscript. LT contributed to interpretation of the radiologic findings and manuscript revision. SC contributed to the interpretation of the genetic findings and manuscript revision. HA, LM, DAH, HH, IL, ZJ, AVK, and SS contributed clinical expertise and critically revised the manuscript for important intellectual content. DMC supervised the project, contributed to data interpretation, and critically revised the manuscript. All authors reviewed and approved the final manuscript.

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Use of large language models

AI assisted in formatting journal requirements, and reference verification; it was not used to generate clinical data or scientific interpretations, and the authors take full responsibility for the content.

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Figures

Figure 1A

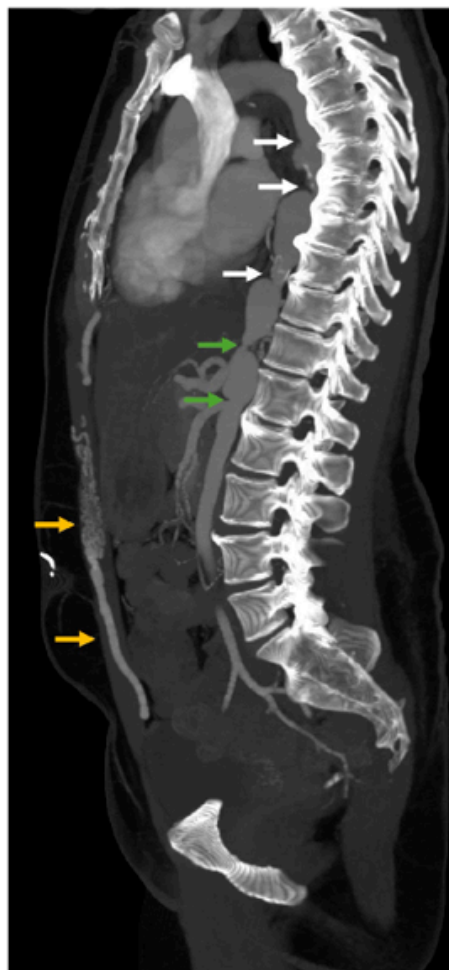


Figure 1B



Figure 1

Thoracoabdominal Aortic Narrowing with Extensive Collateralization on Computed Tomography (CT) Angiography

A: Sagittal CT Angiogram Demonstrating Multifocal Aortic Narrowing (white arrows) and Collateral Vessels Sagittal oblique arterial-phase CT angiogram with maximum intensity projection (MIP) reformat demonstrates multifocal thoracic (white arrows) and abdominal (green arrows) aortic stenosis with post-stenotic ectasia. Dilated collateral vessels in the abdominal wall (orange arrows) contribute to the common femoral arteries (not shown).

B: Three-dimensional reconstruction of the thoracic and upper abdominal aorta highlights the multifocal stenosis and post-stenotic ectasia.

Neurological:
Ischemic stroke

Developmental:
Learning difficulties, requires special education

Craniofacial:
Sparse eyebrows, short neck

Cardiovascular:
Malignant hypertension;
thoracoabdominal aortic
vasculopathy with multifocal
narrowing and collateral
circulation

Renal:
Solitary right kidney (left
nephrectomy in infancy due
to hypoplastic kidney)

Family History:
Father: kidney failure, stroke
Brother: developmental delay

Additional findings: Poorly rooted teeth, Decreased mandibular bone density

Affected Homolog (Deletion)

Unaffected Homolog

22q11.21 Deletion

954.8 kb Heterozygous Deletion, chr22:20,459,997-21,414,845 (GRCh37/hg19)

LCR22B

SCARF2

PI4KA

SERPIND1

SNAP29

CRKL

LZTR1

LCR22D

Centromere

p

q

q11.1

q11.21

q11.23

q12

q13.3

q

q11.1

q11.21

q11.23

q12

q13.3

0 Mb

Clinical Phenotype of Central 22q11.2 Deletion

Page 13/14

B: Schematic ideogram of chromosome 22 showing the affected homolog (left) and the unaffected homolog (right). The affected homolog harbors a heterozygous deletion at band 22q11.21, spanning approximately 954.8 kb (chr22:20,459,997–21,414,845; GRCh37/hg19). The deleted interval is delineated by the flanking low-copy repeats LCR22B (proximal) and LCR22D (distal), consistent with a recombination-mediated rearrangement. Chromosomal bands, centromere position, and p and q arms are indicated for orientation. The unaffected homolog demonstrates an intact 22q11.2 region.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- [tableS2geneswithindeletion.pdf](#)
- [TableS1CAKUTgenesfinal29072026.pdf](#)