



Hospitais da Universidade de Coimbra  
Clínica Universitária de Imagiologia  
Prof. Doutor Filipe Caseiro Alves

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# Caso Clínico

28/09/2011

Maria João Magalhães



# Informação Clínica

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## Identificação

- M.A.R.C.F
- Sexo feminino
- 54 anos
- Caucasiana



# Informação Clínica

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## História Clínica

- Distensão abdominal e enfartamento pós-prandial
- Sem perda de peso, vômitos ou obstipação

AP: Colecistectomia (2000), amigdalectomia, osteoporose

AF: Irrelevantes



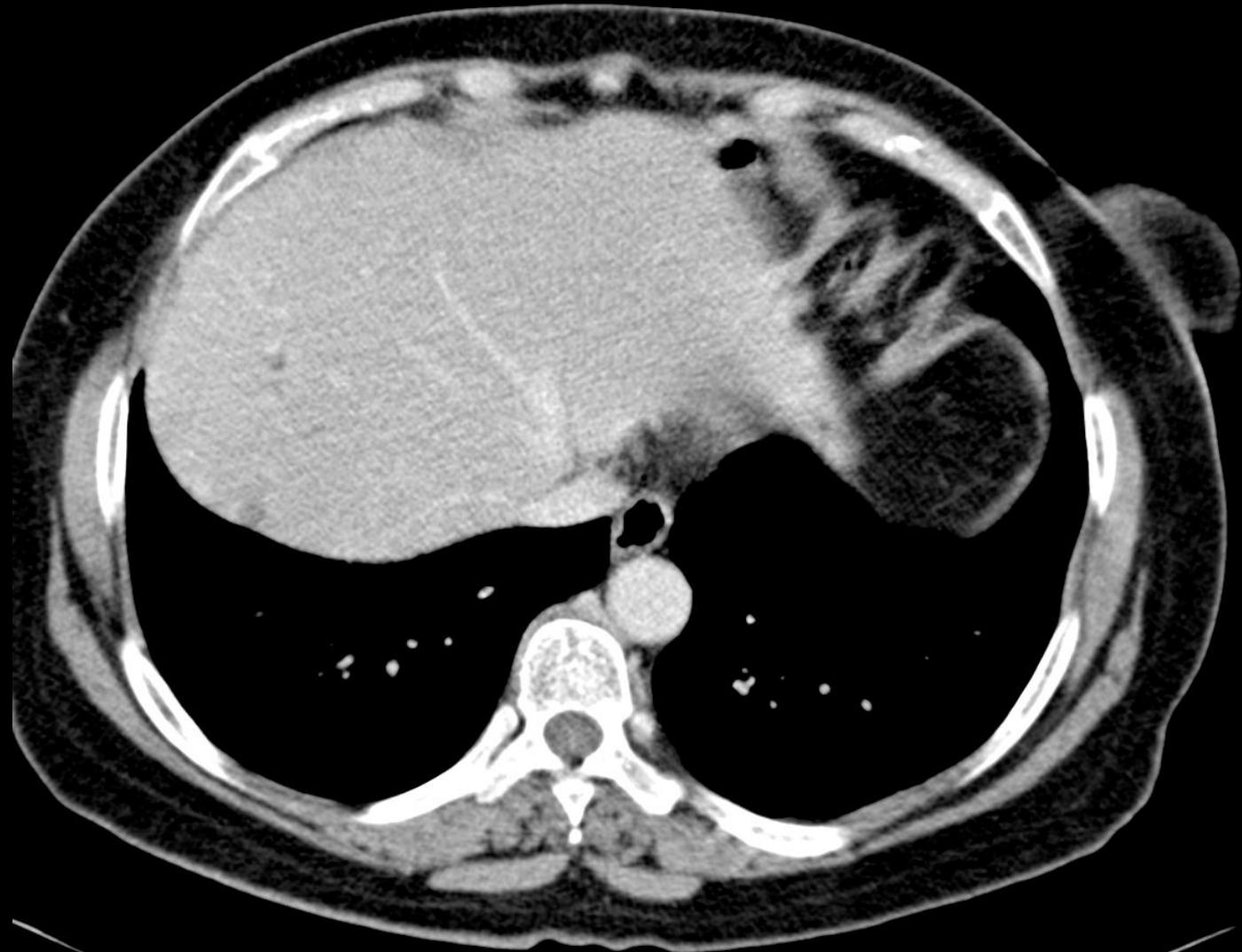
# Informação Clínica

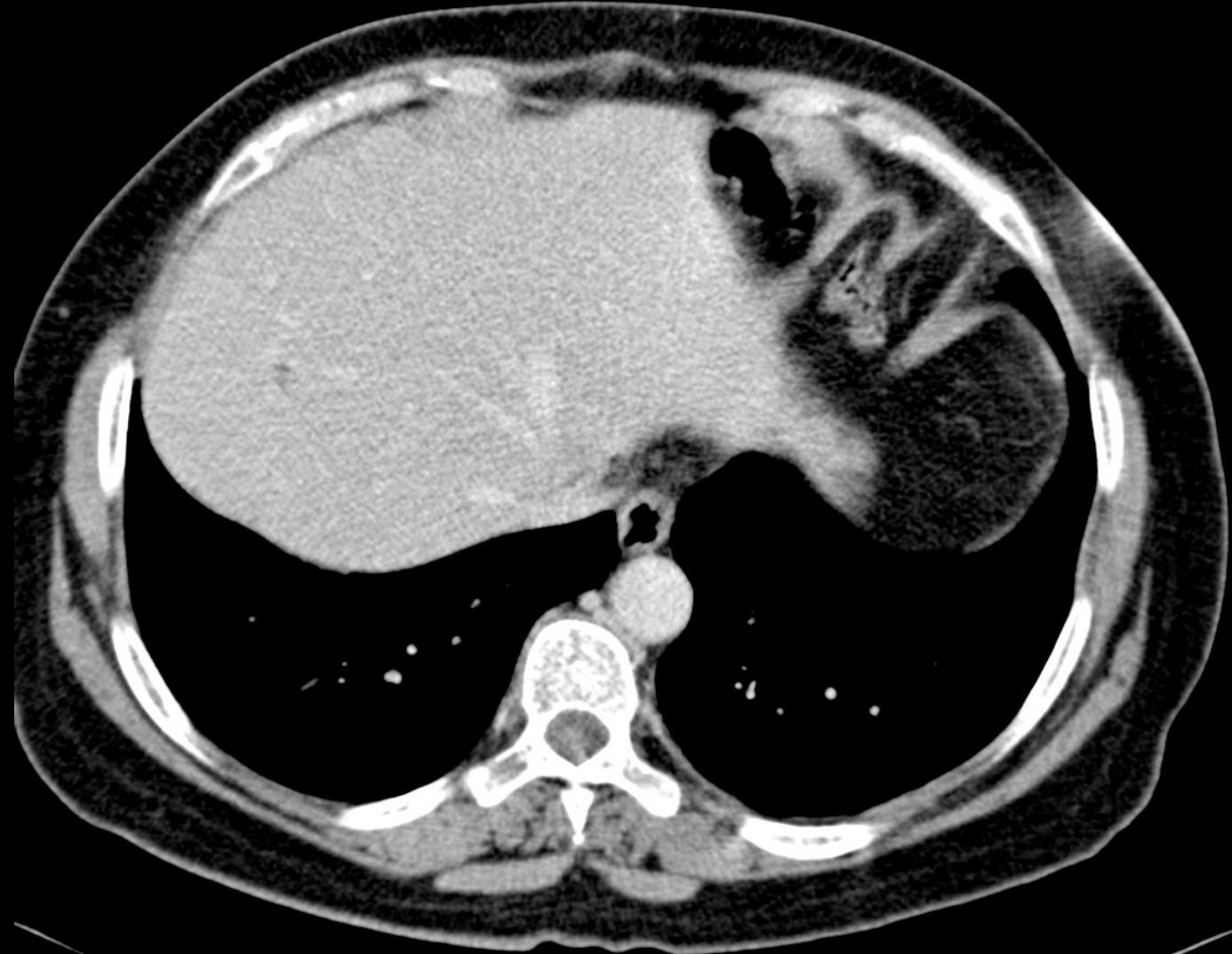
- AngioTC → Dilatação vias biliares IH e da VBP (1,1cm)  
(2008) → Espessamento parietal irregular duodeno-jejunal  
→ Lipoma jejunum (14mm)  
→ Densificação gordura mesentérica  
→ Múltiplas formações ganglionares mesentéricas  
→ Formação nodular hipodensa (3x2cm) processo uncinado pâncreas
- Biópsia de nódulo pancreático: “aspectos sugestivos de lesão benigna- pancreatite crónica”
- Biópsia duodenal: “infiltrado inflamatório”

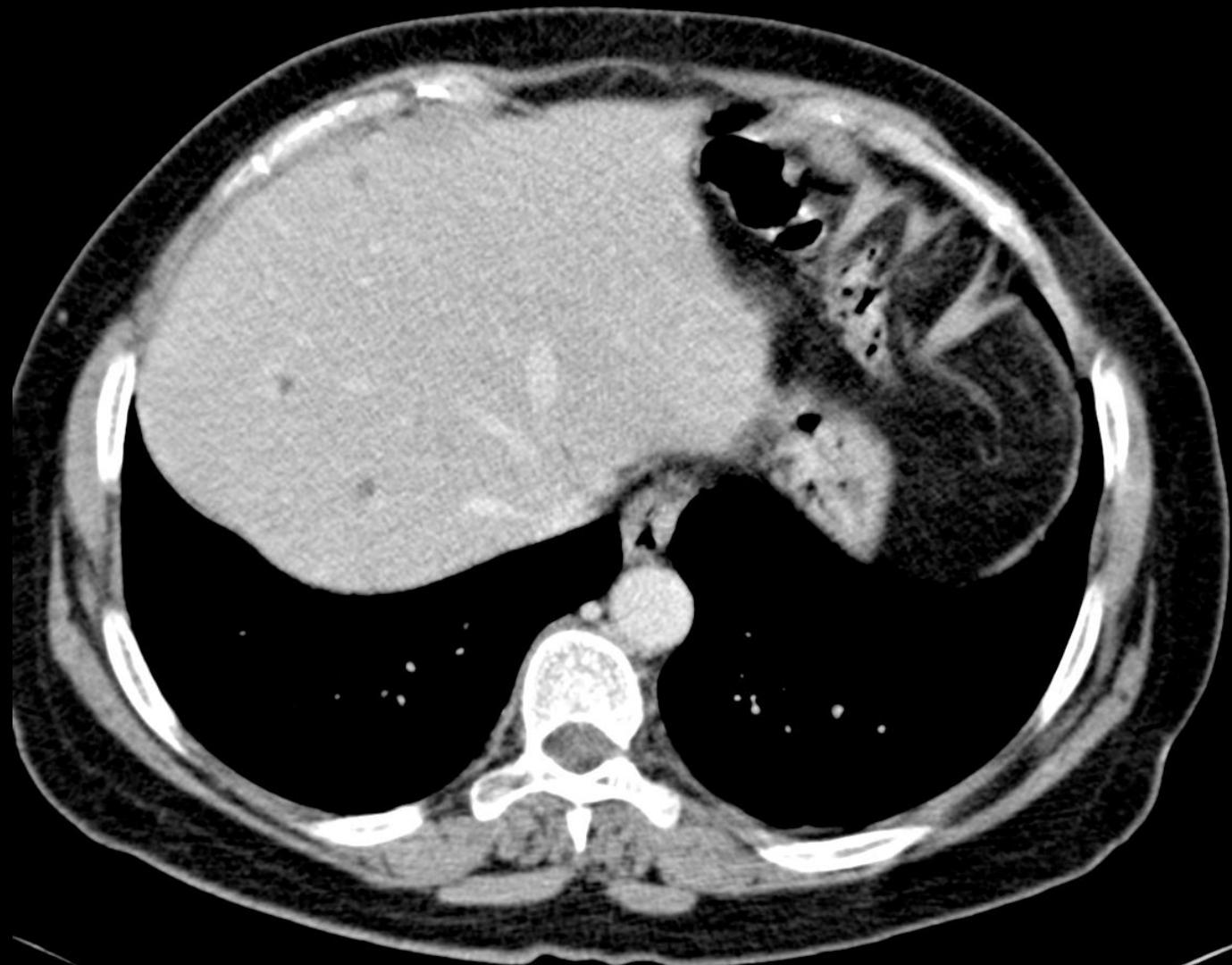


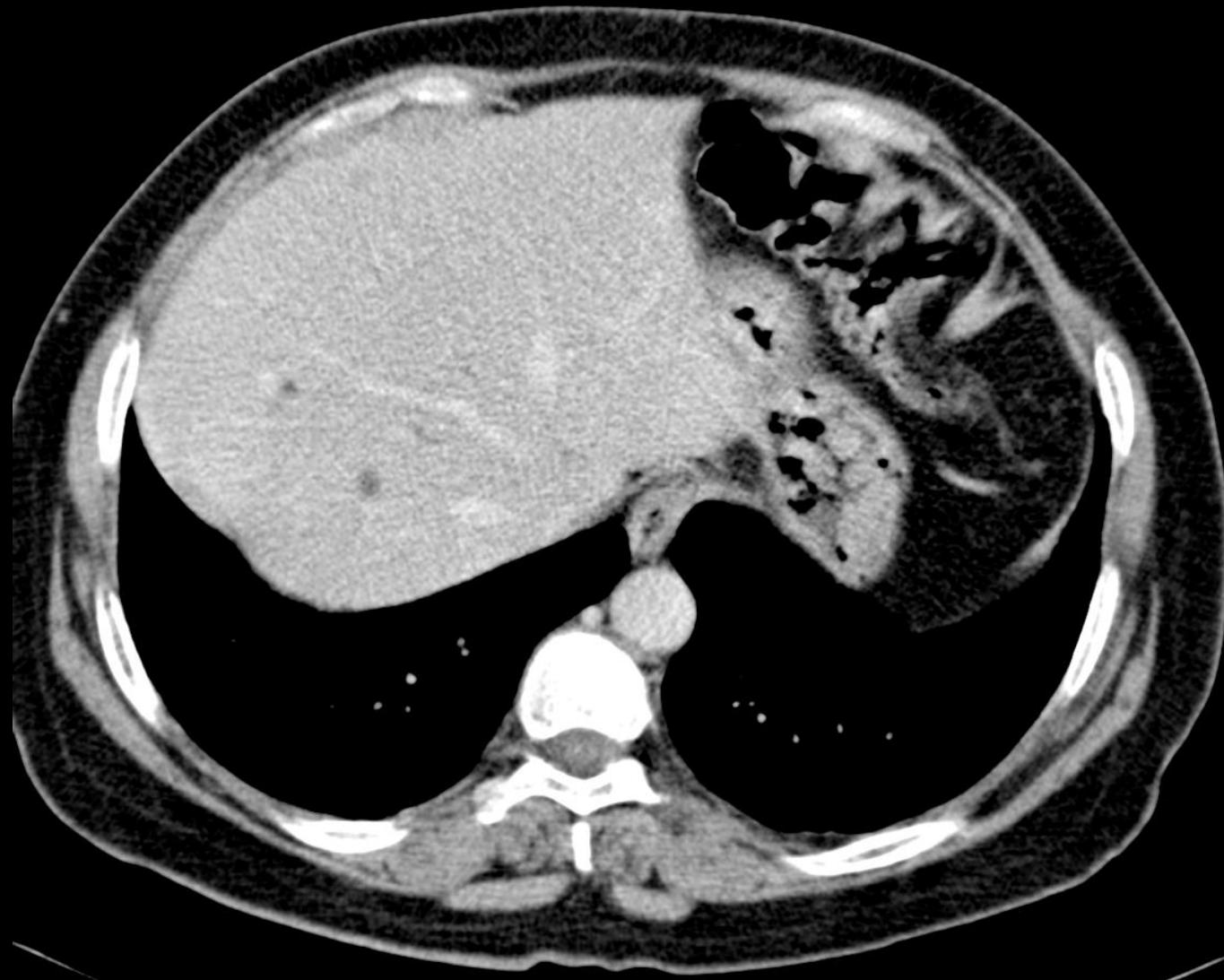
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TC abdominal superior





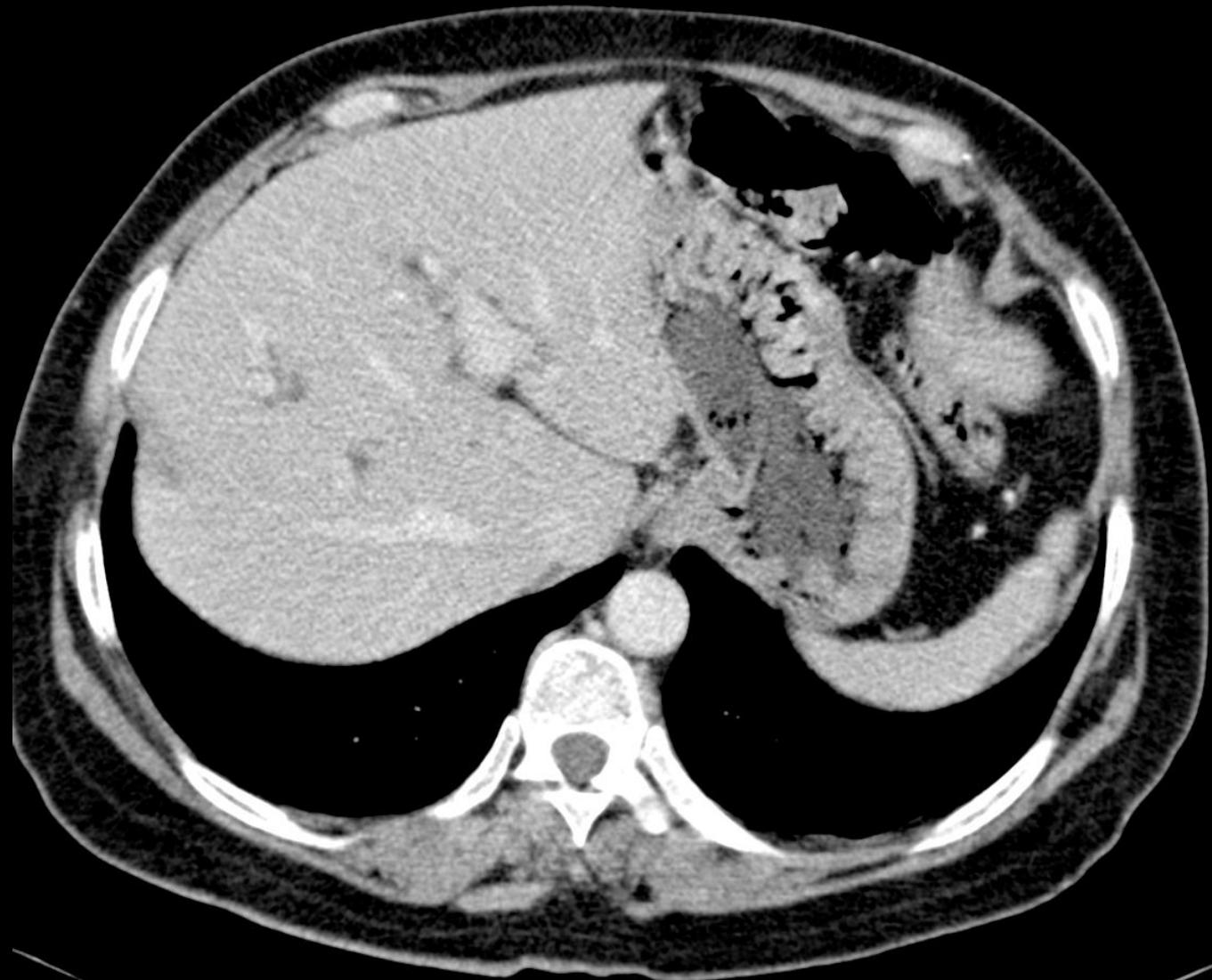




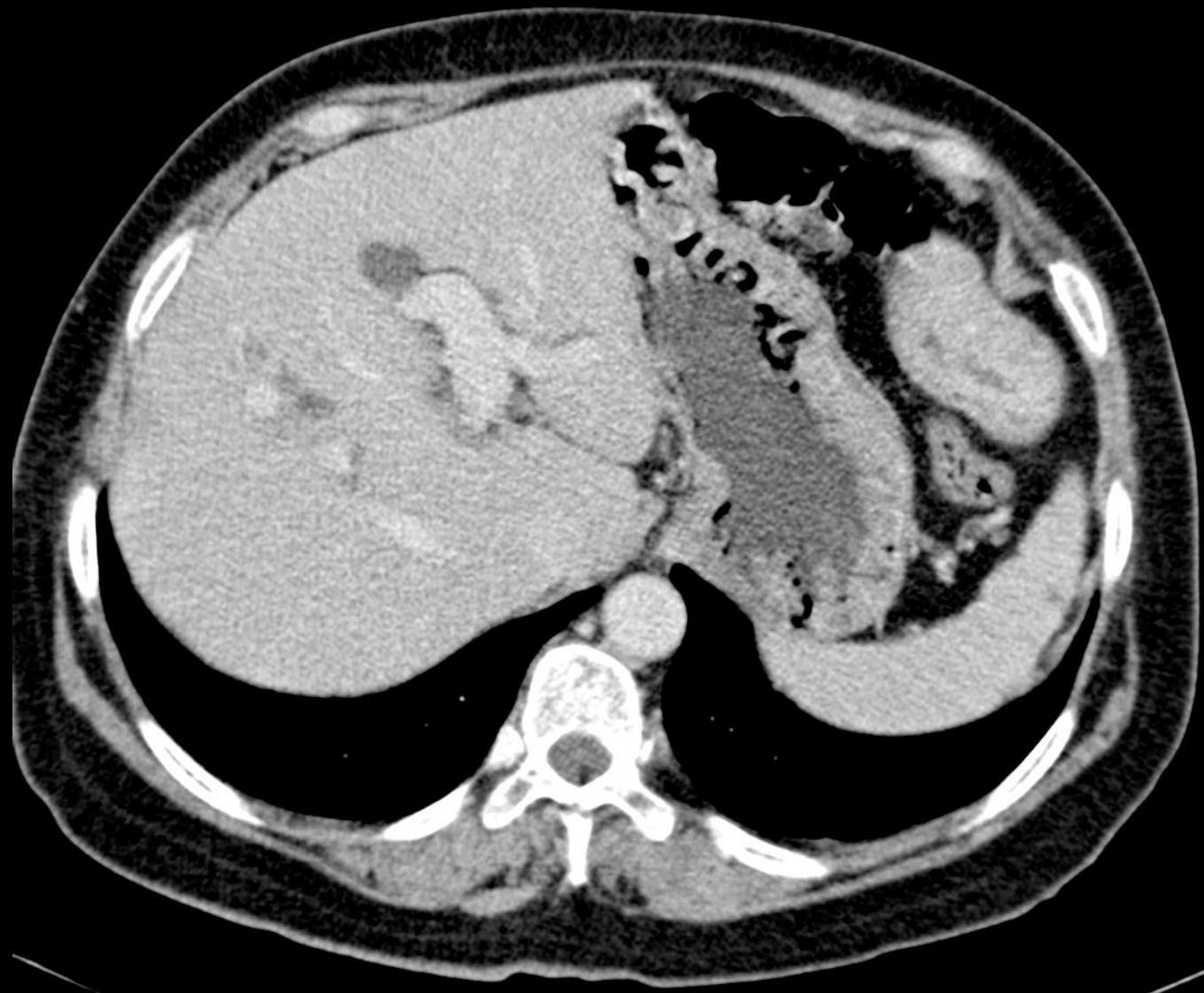




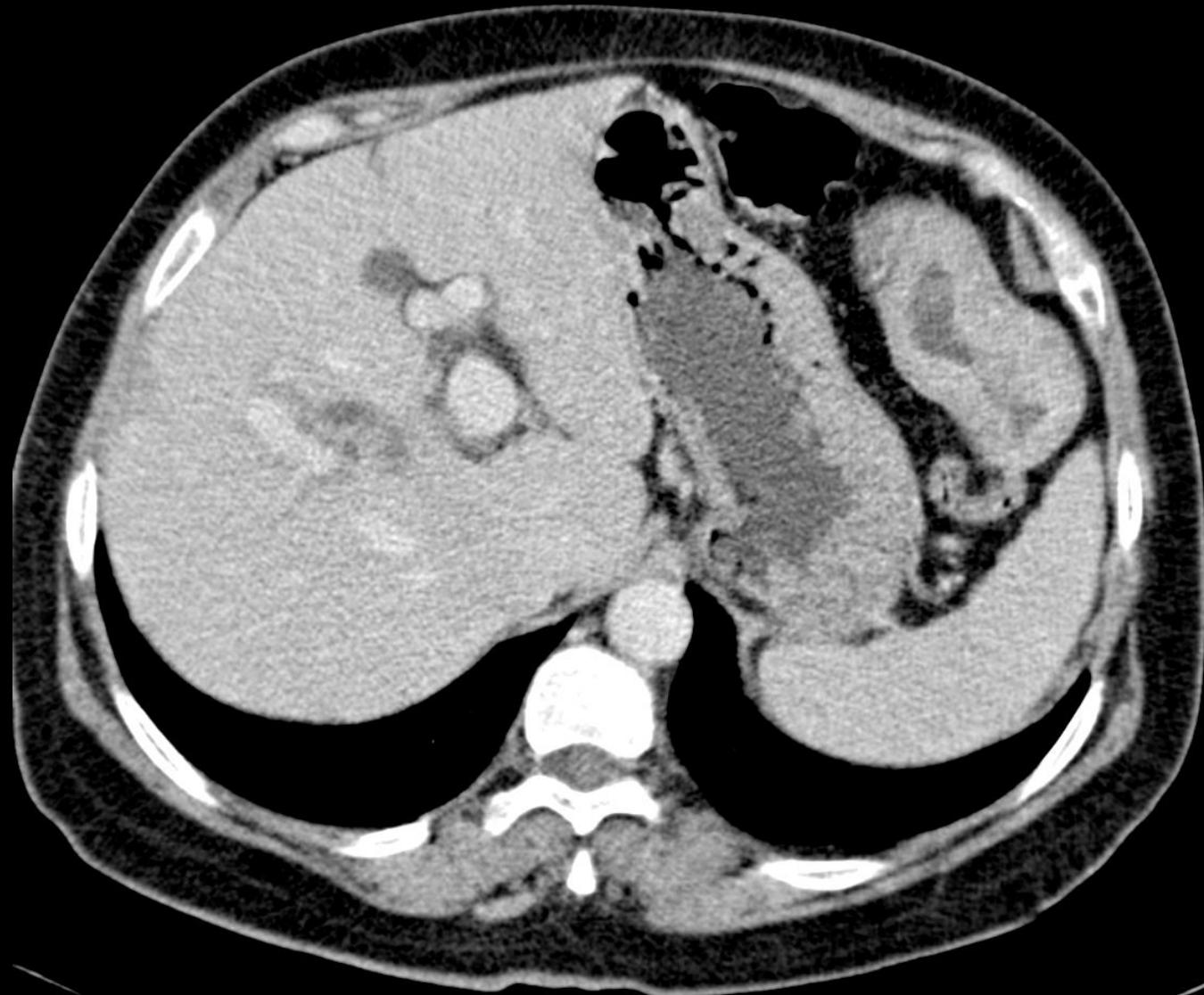


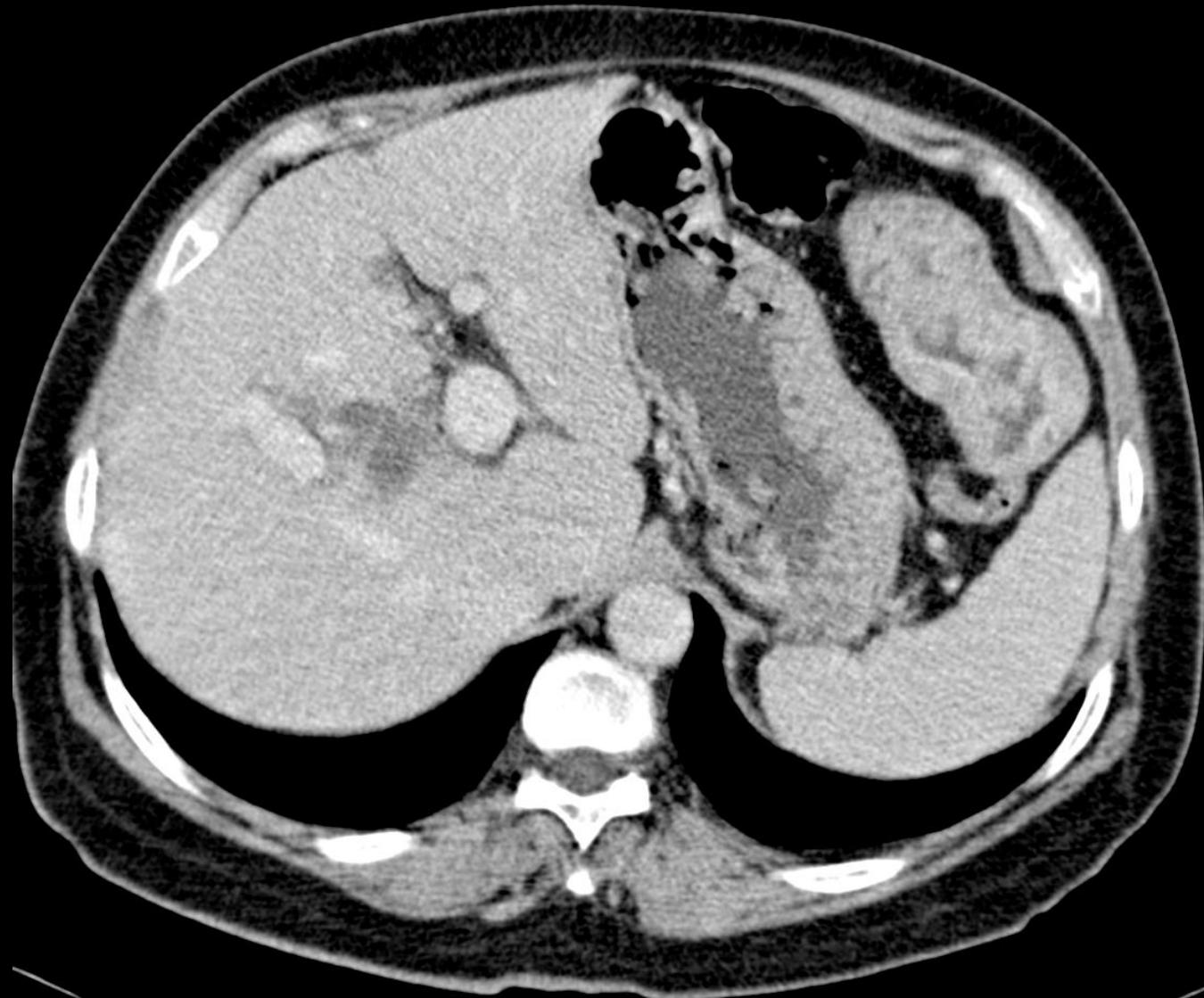


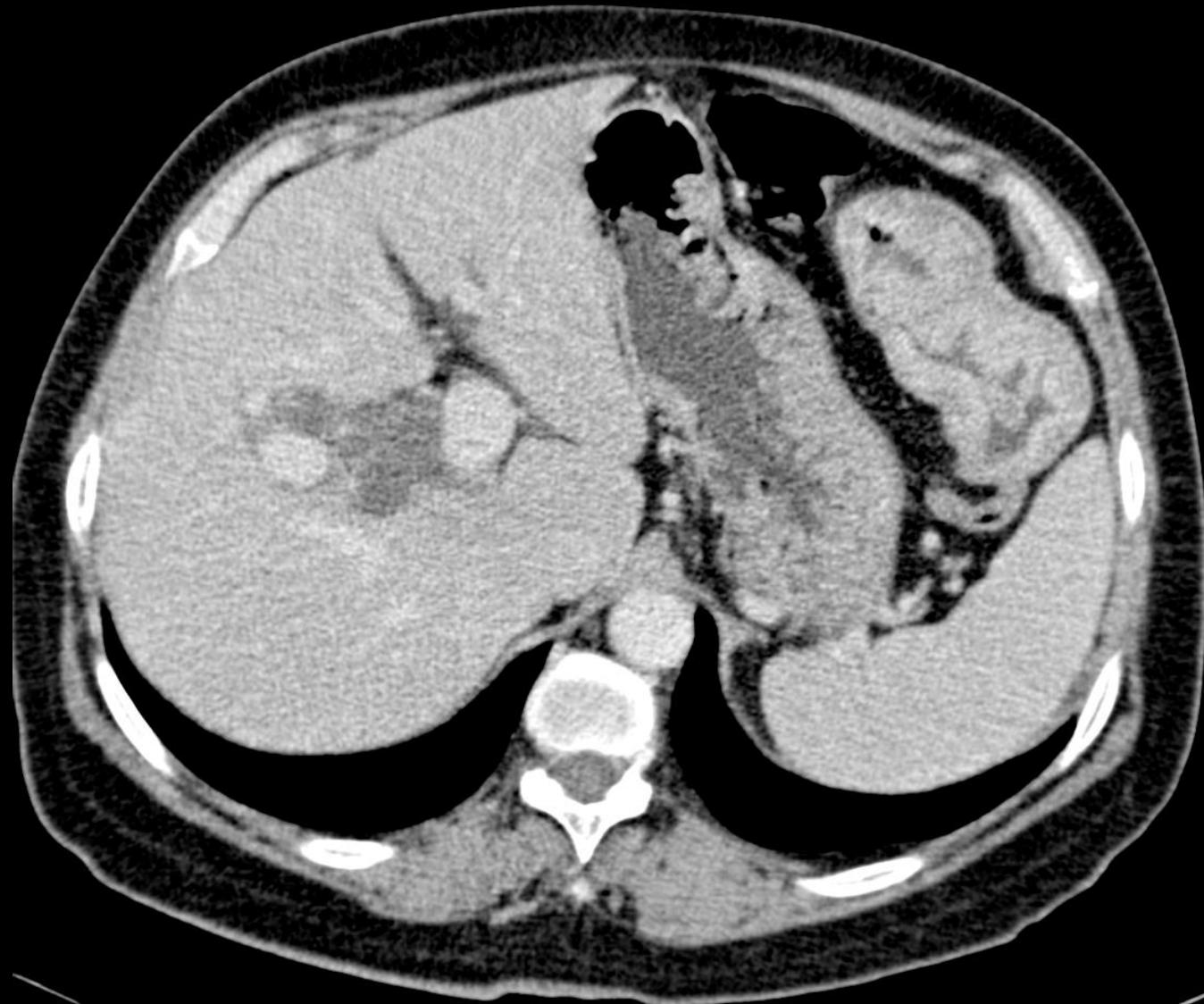


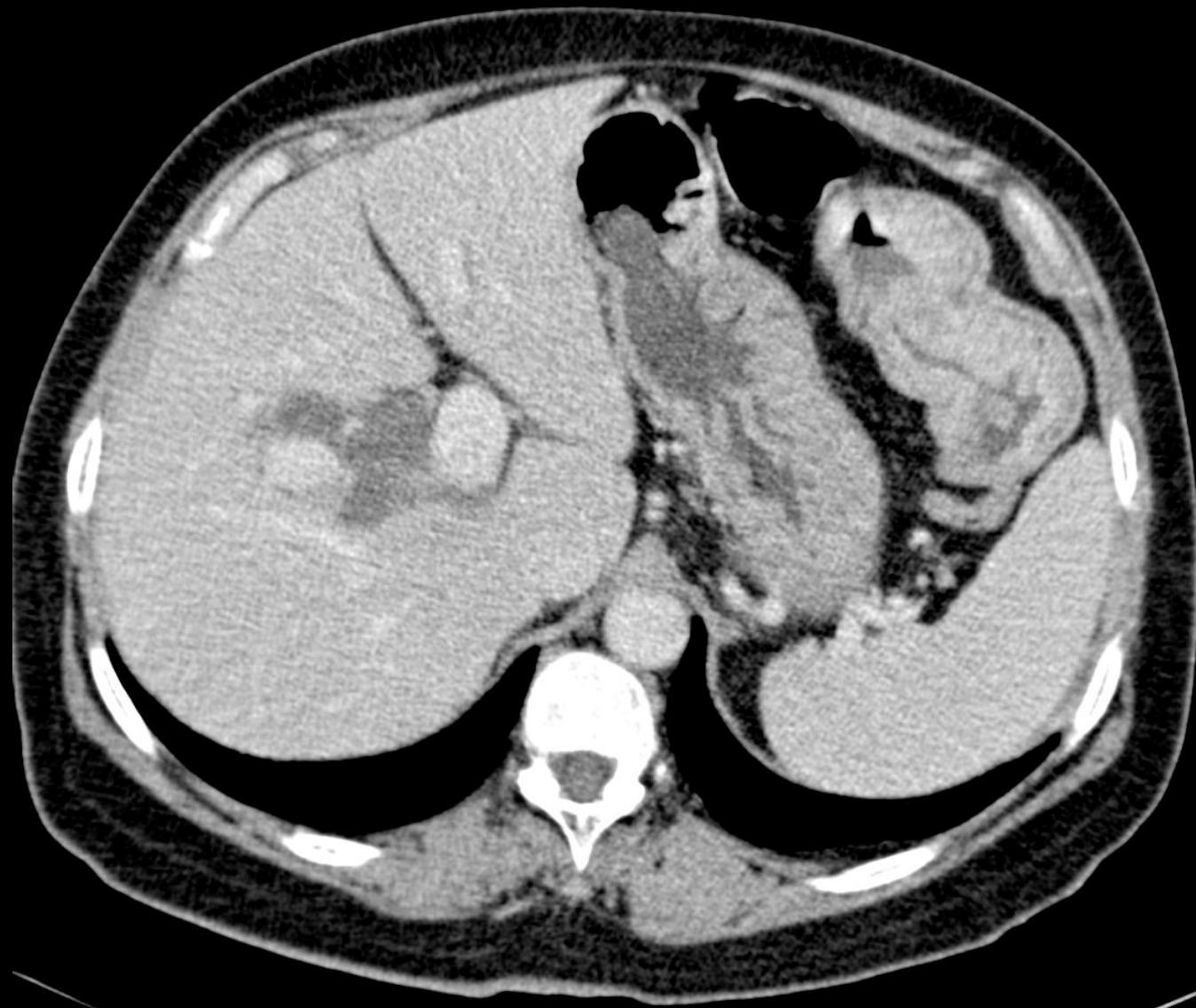


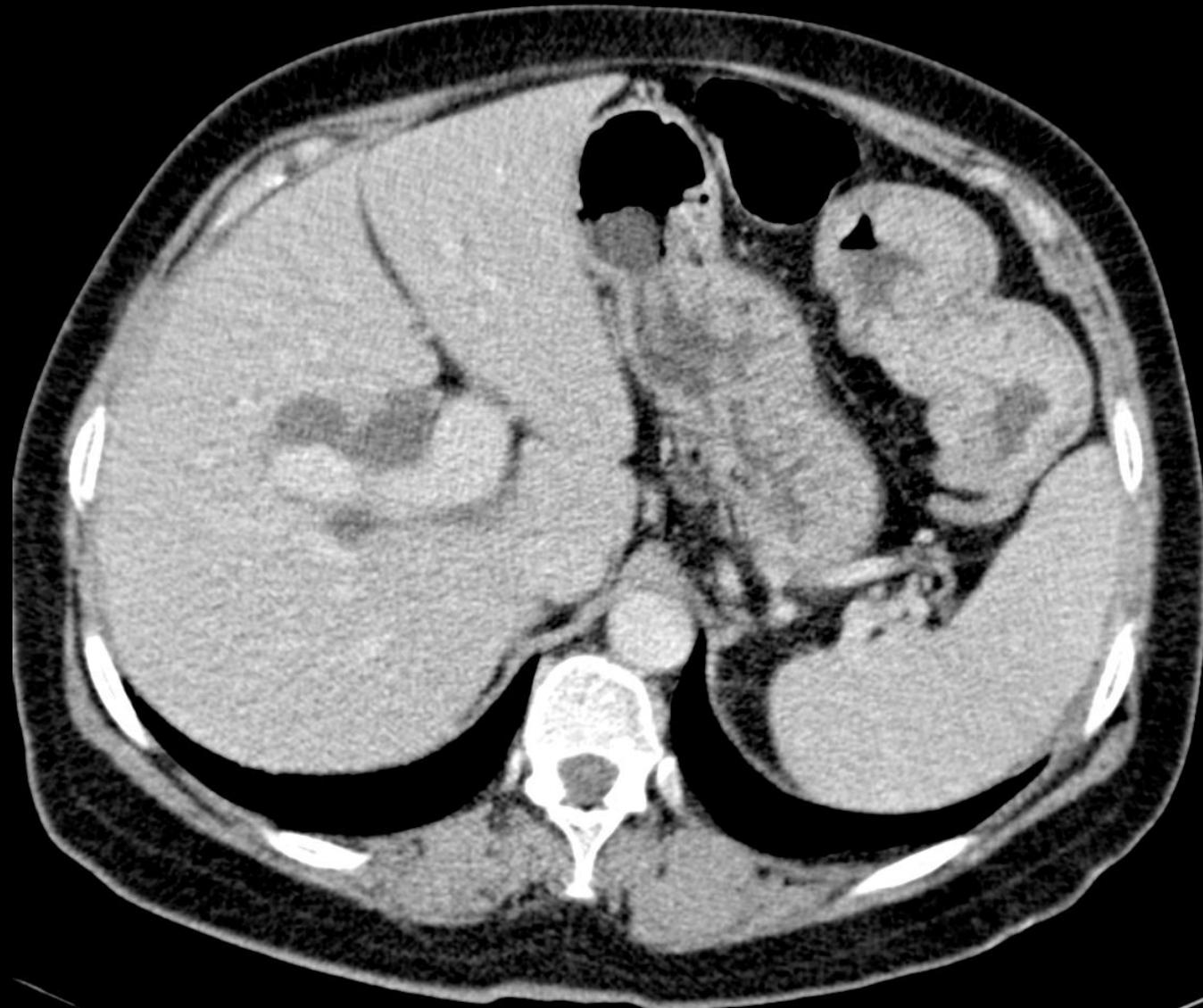


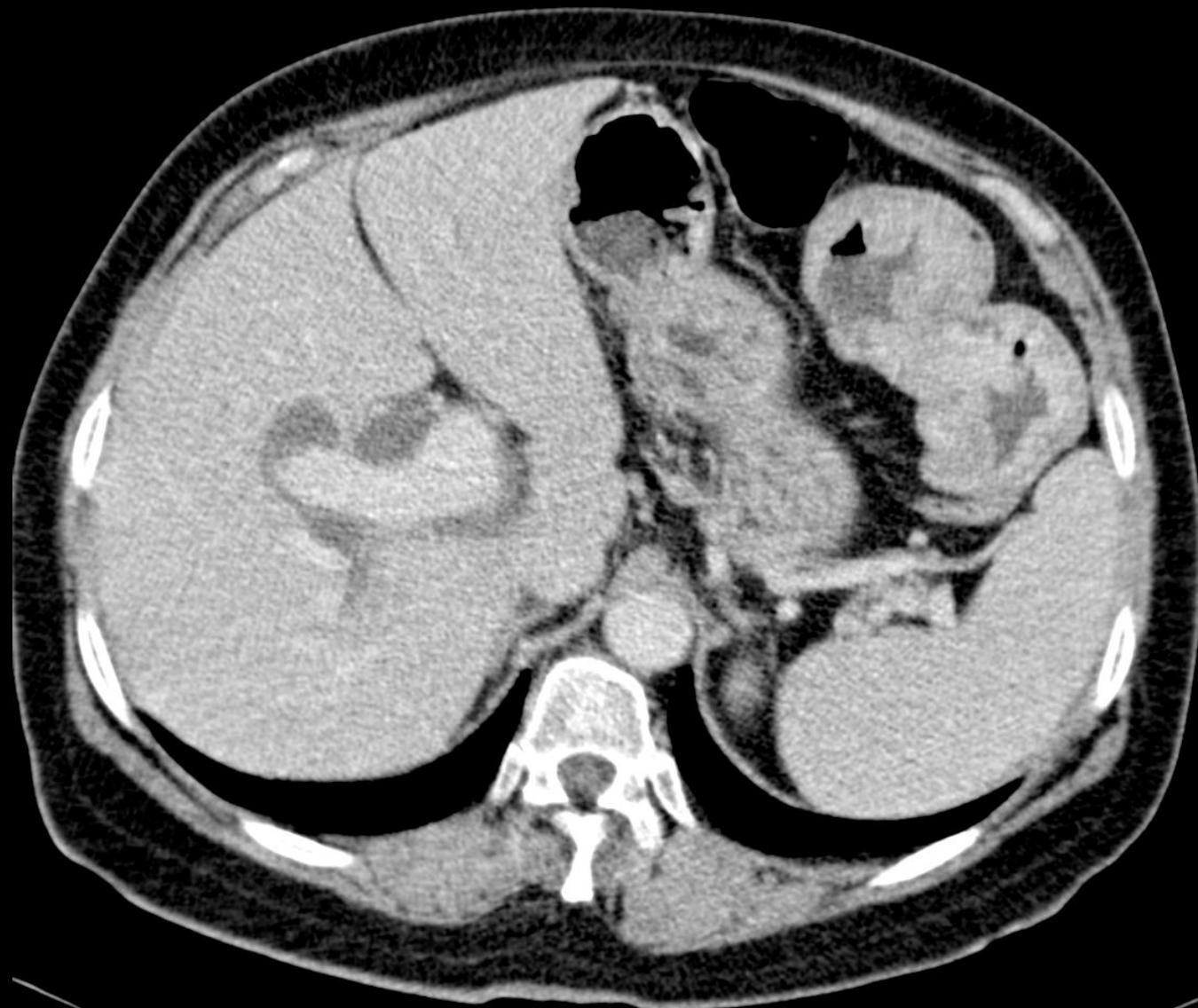


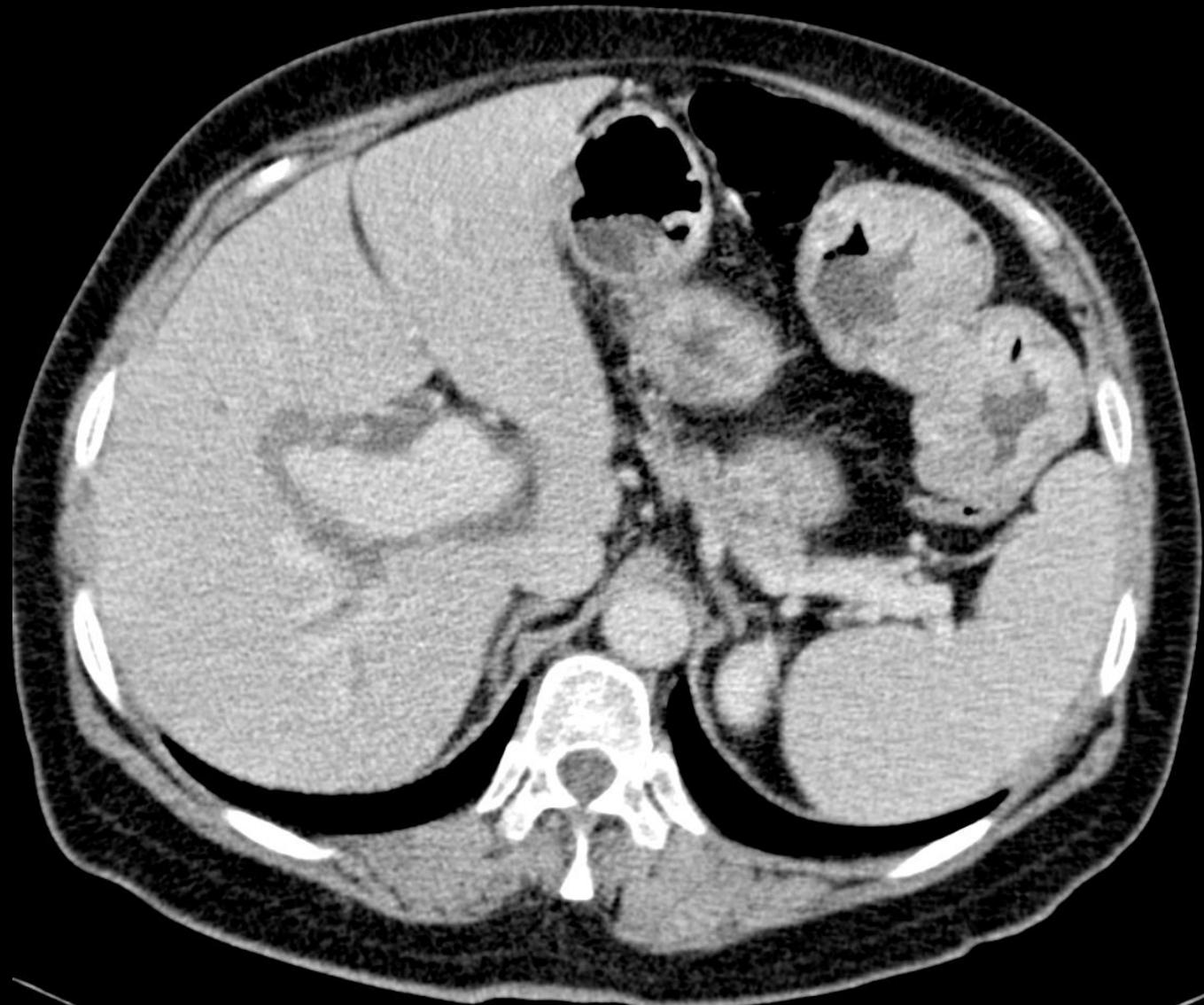


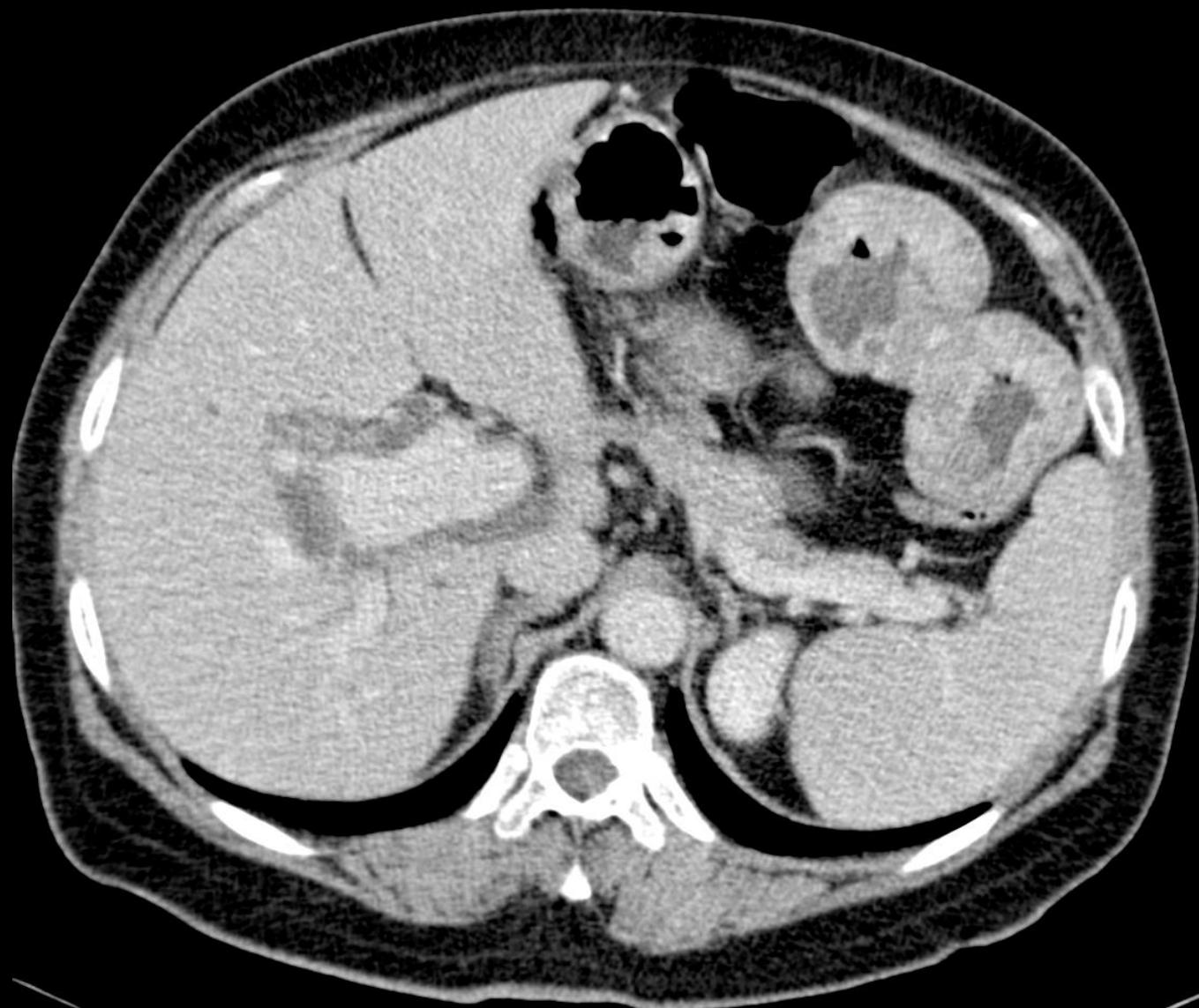


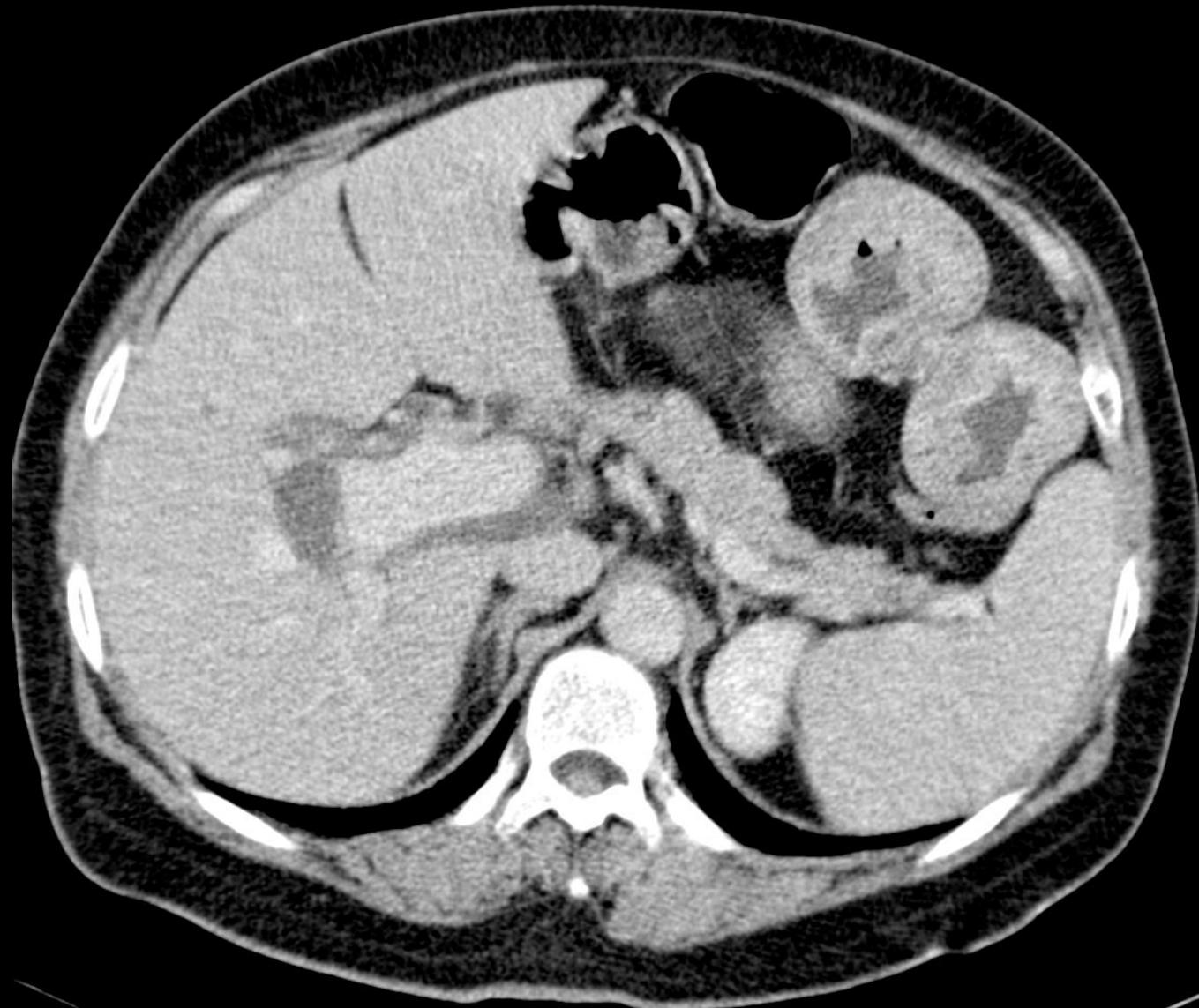


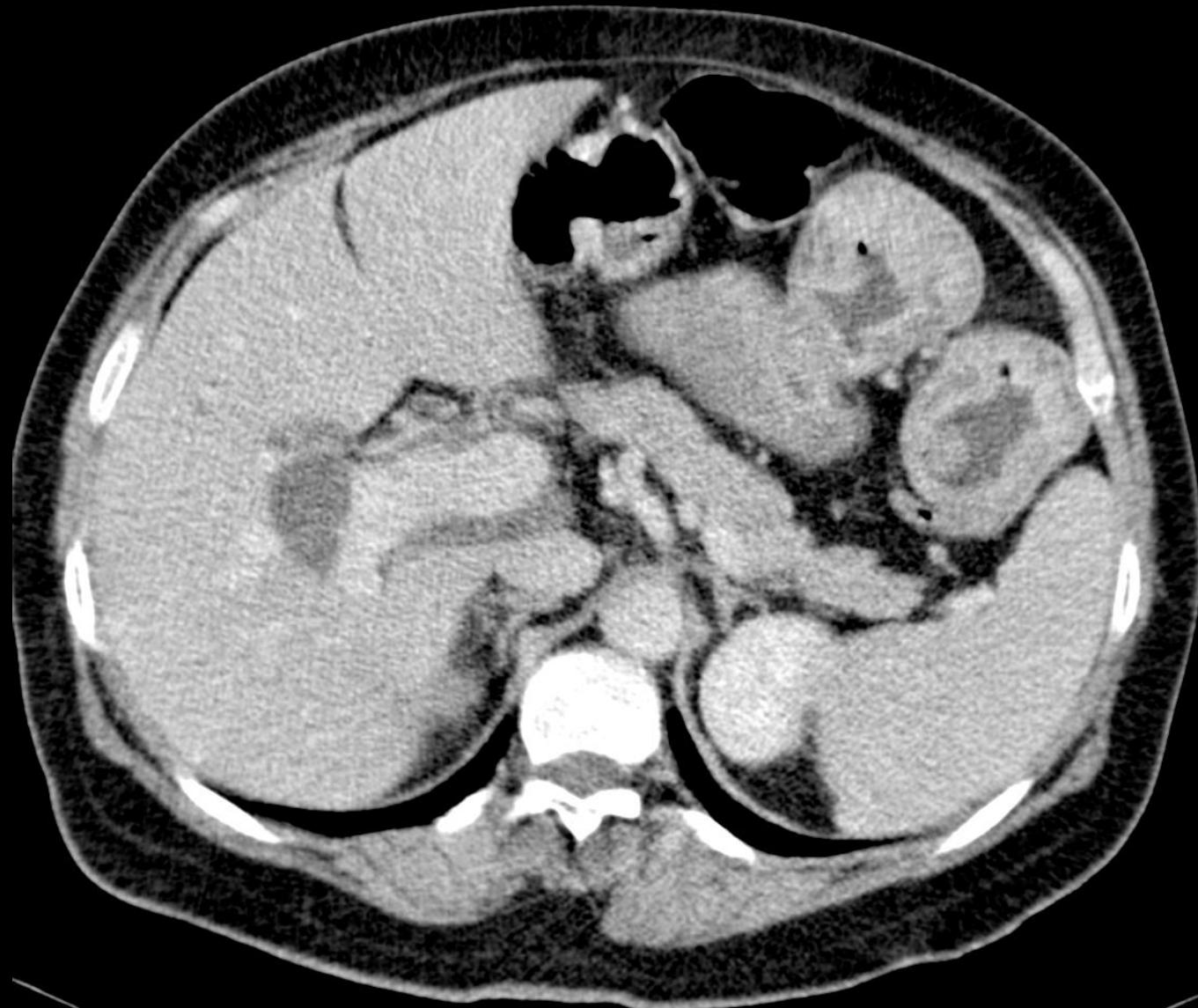


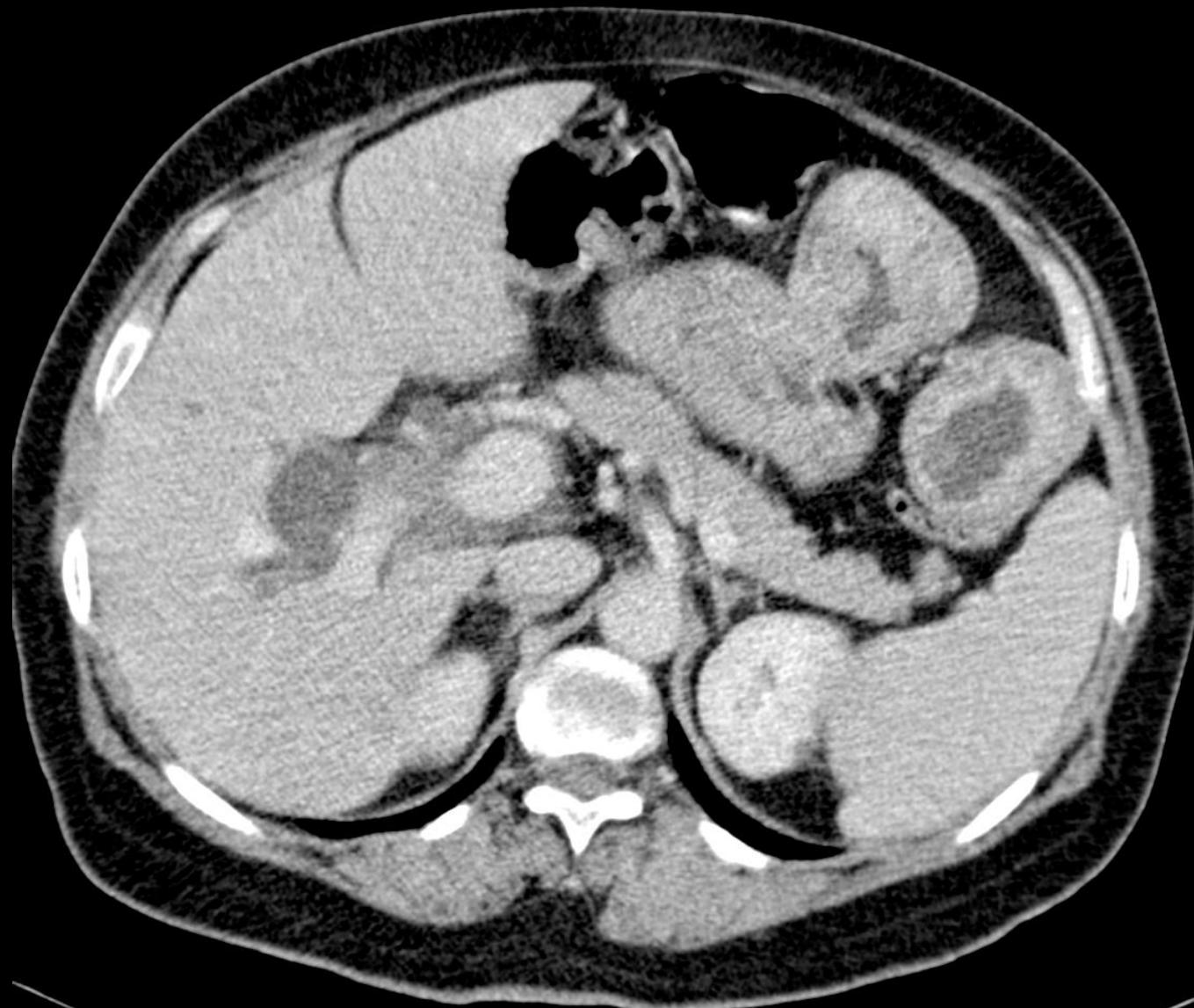


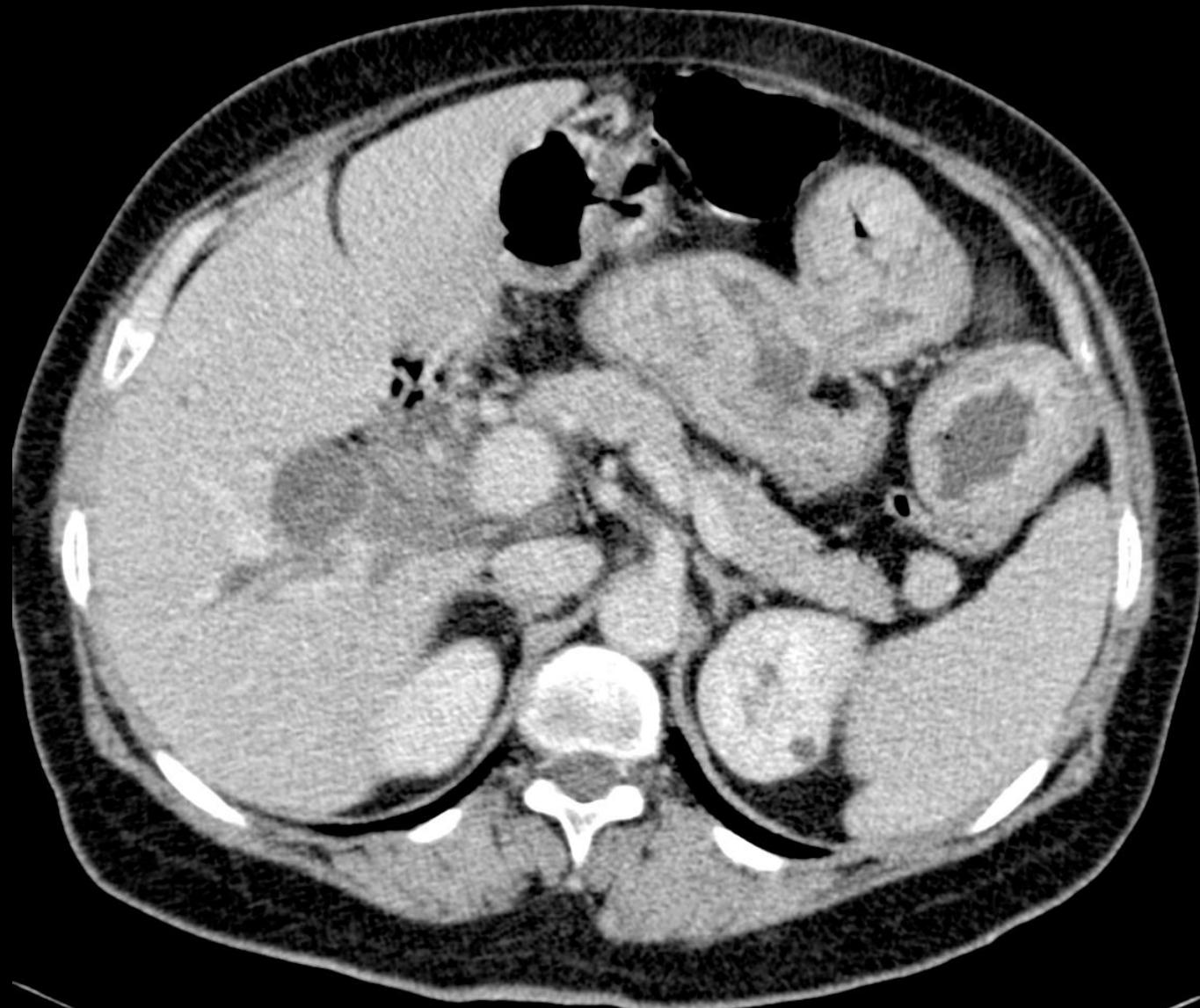


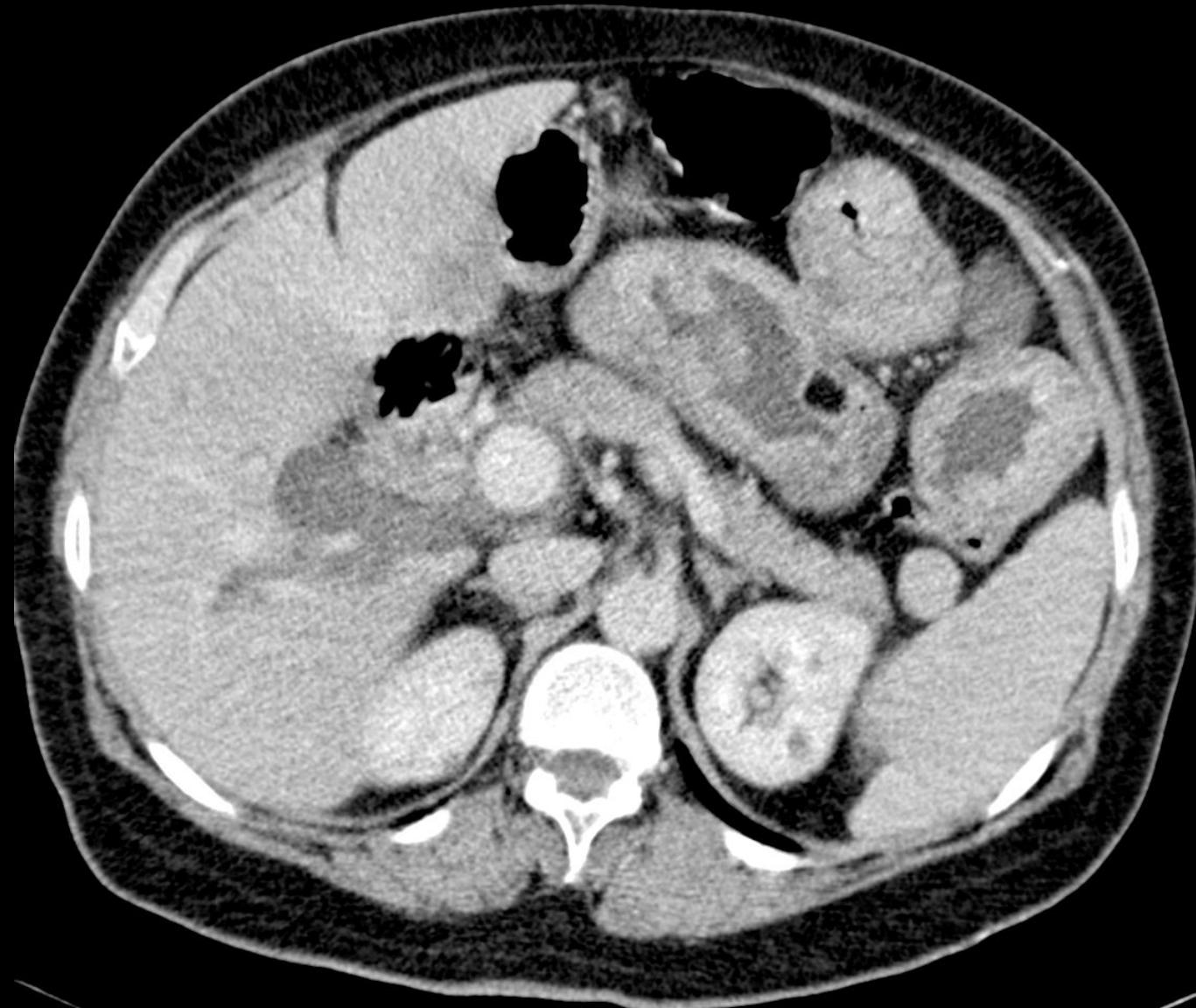


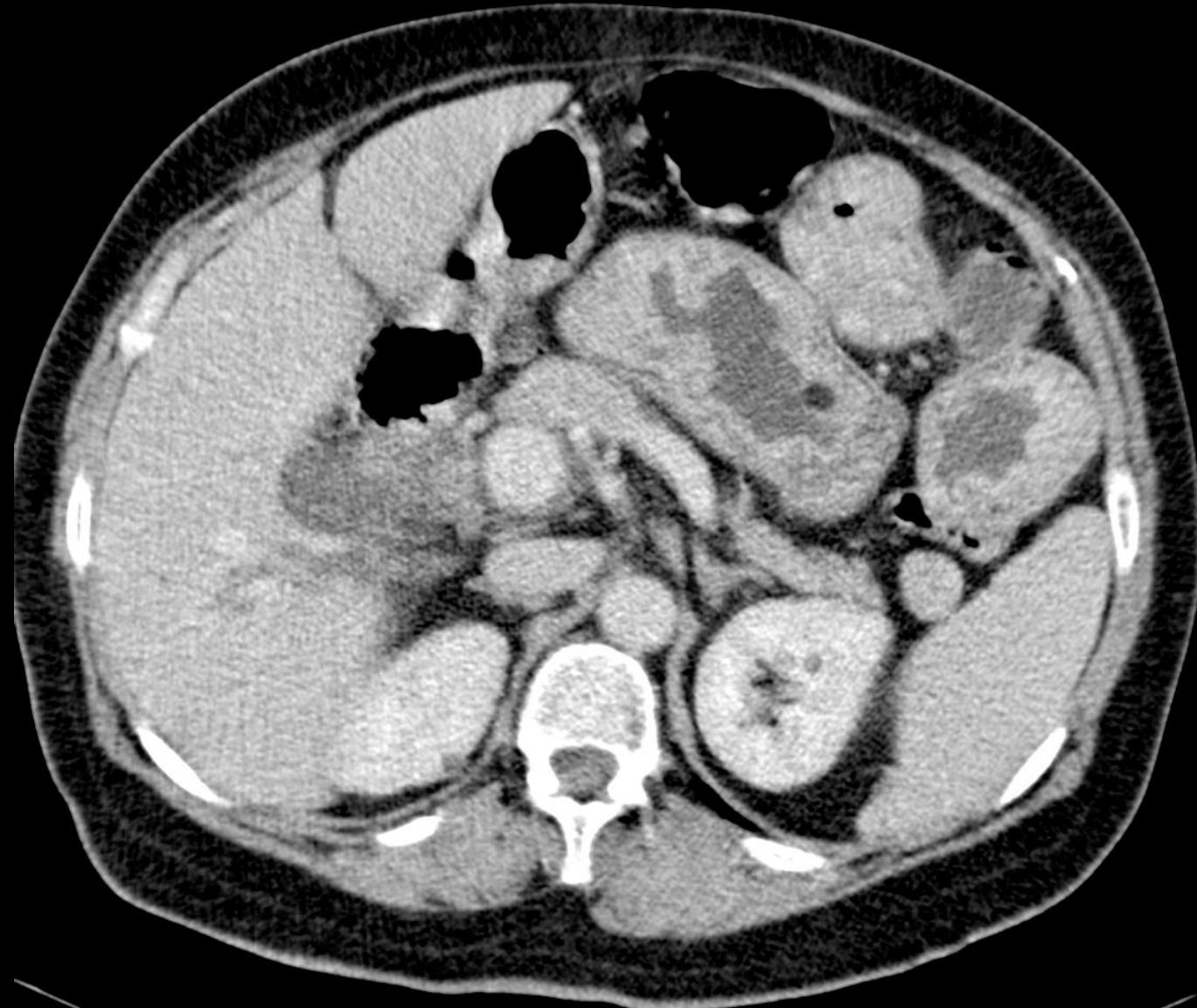


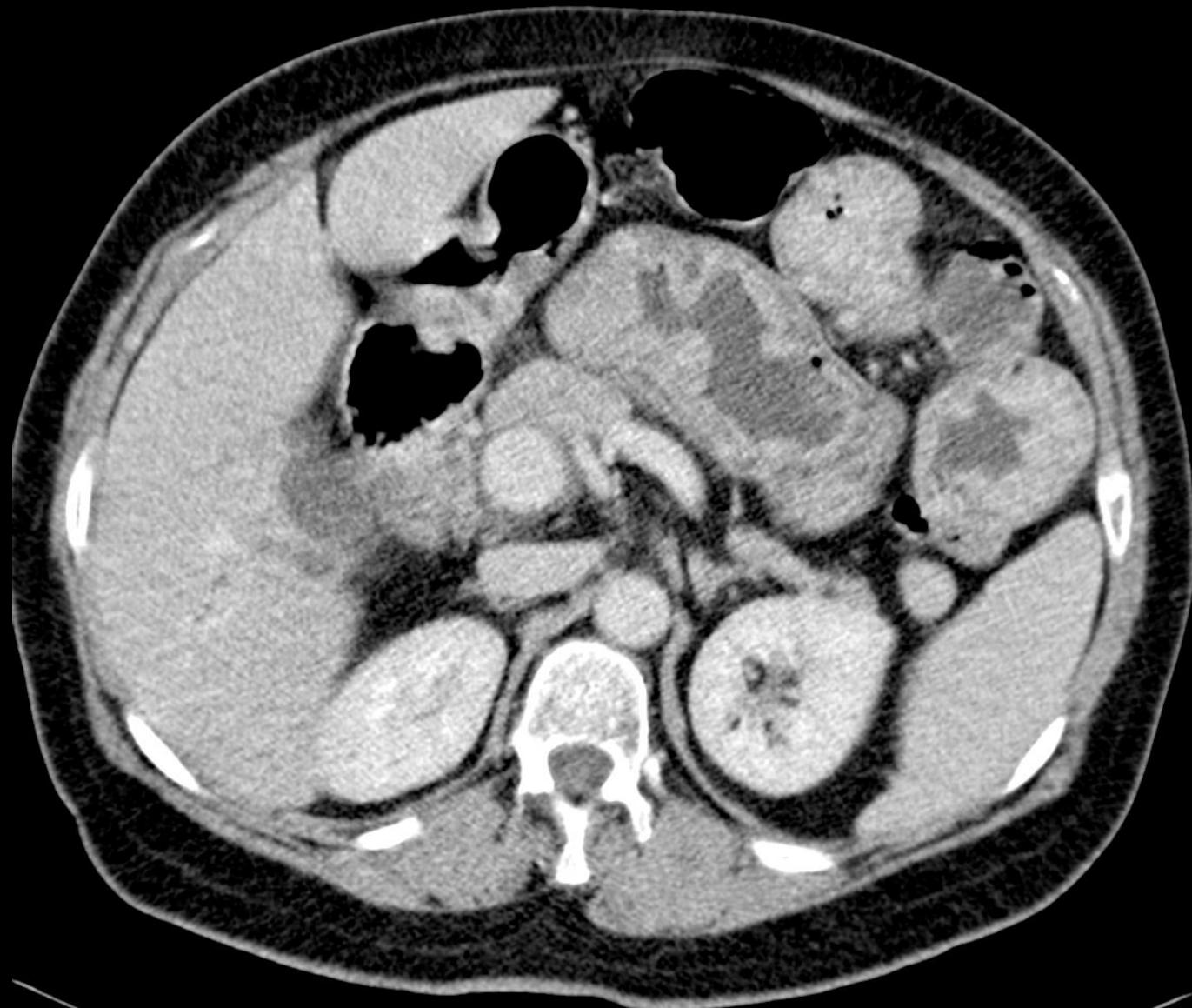


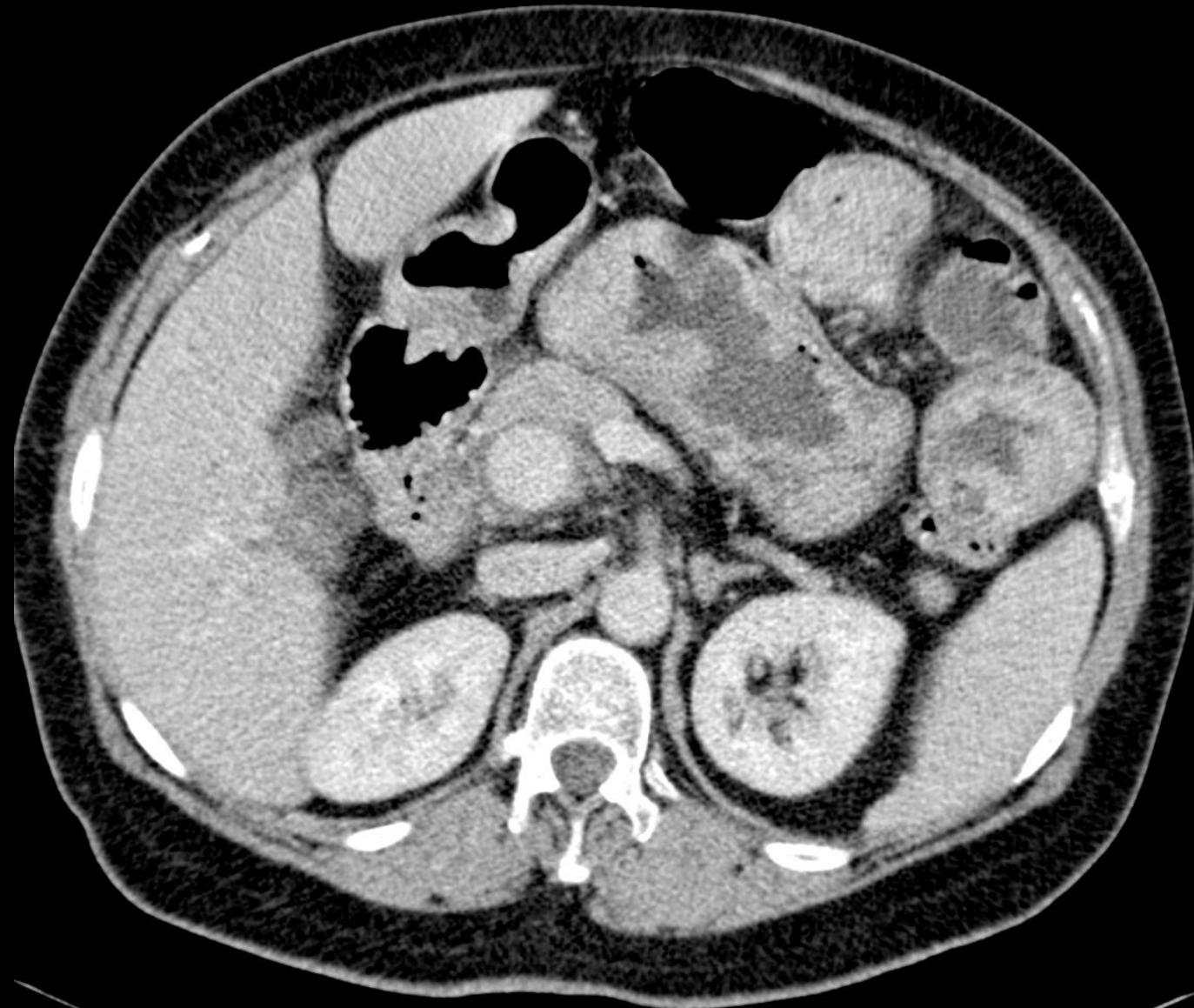


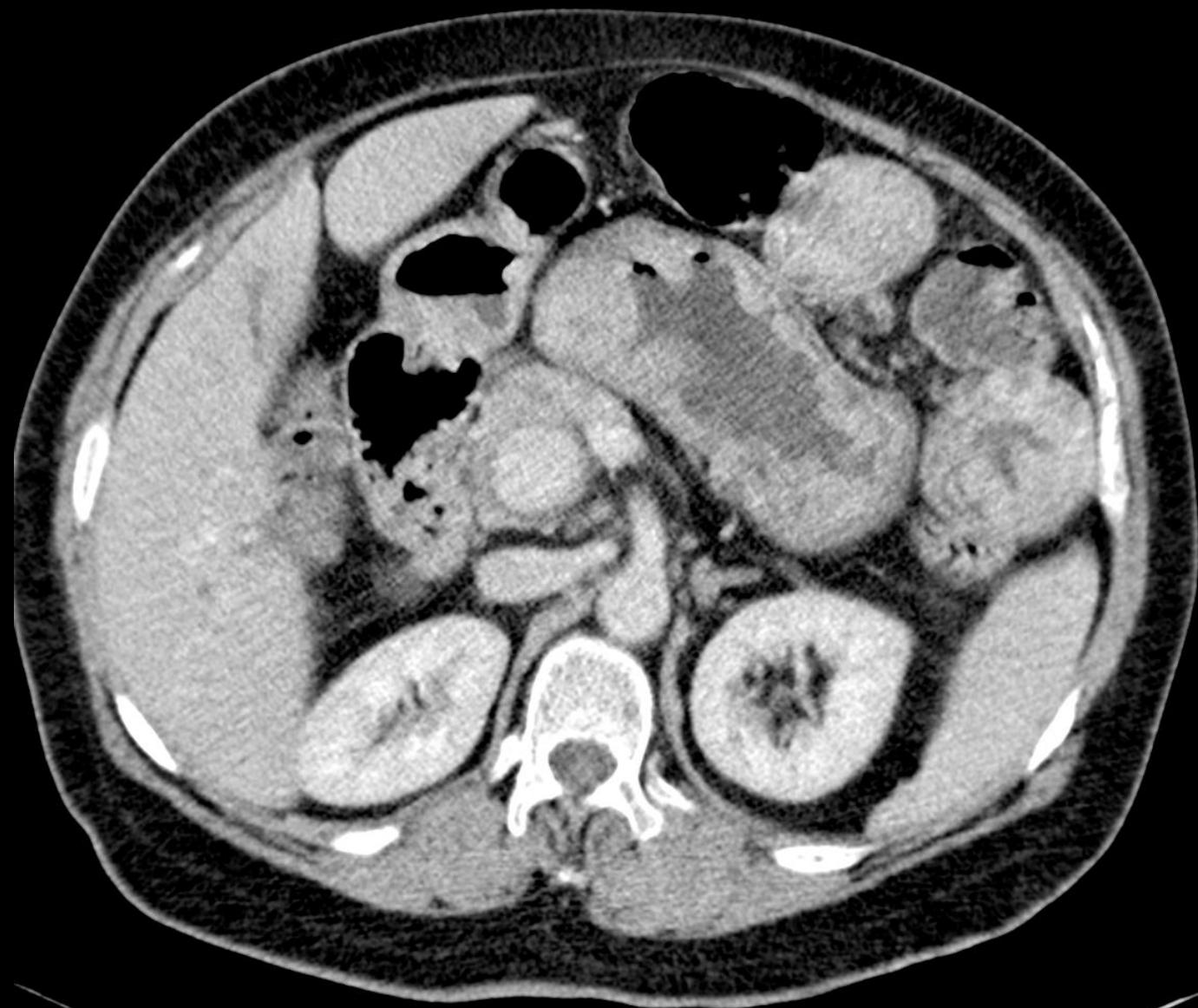


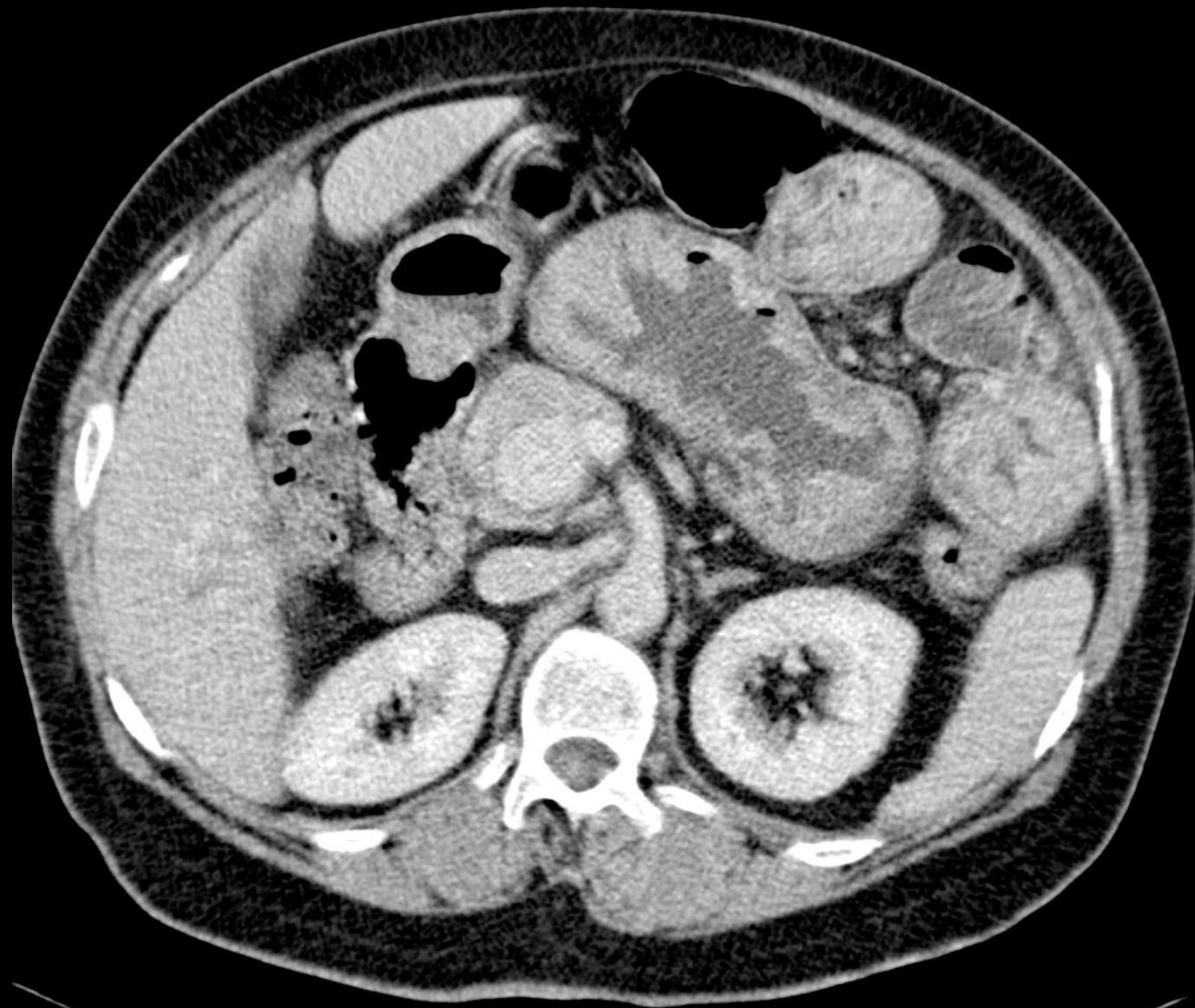


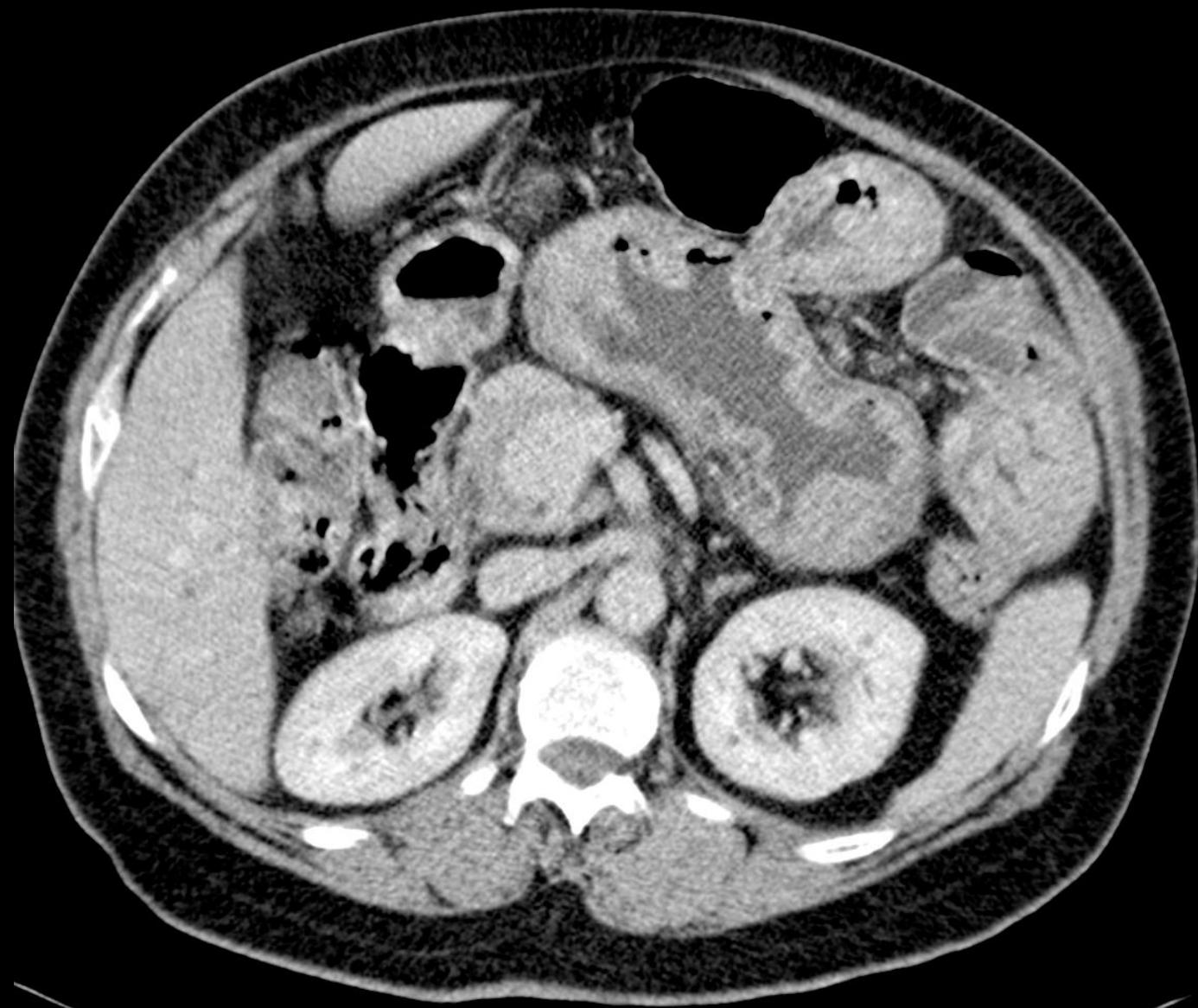


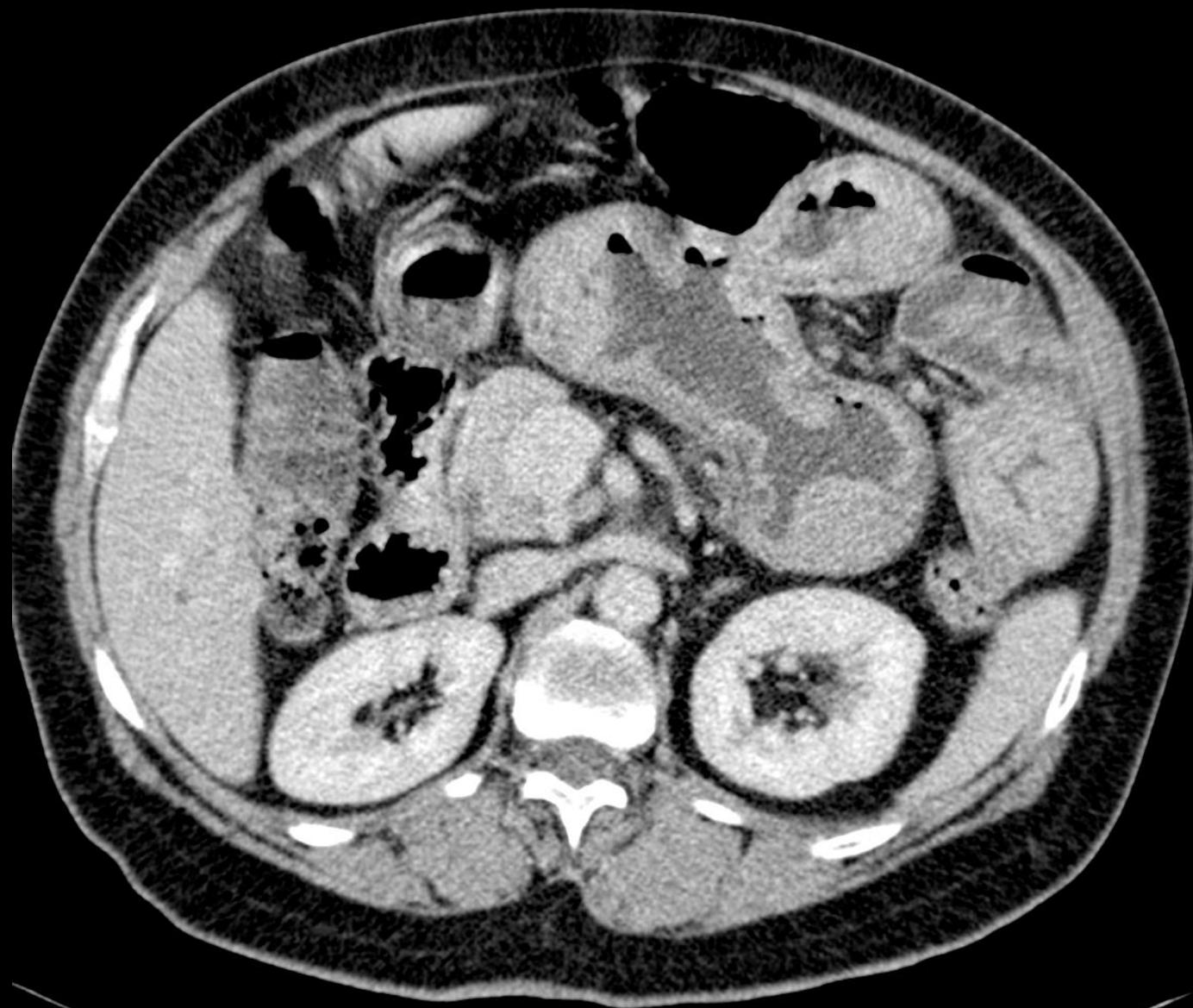


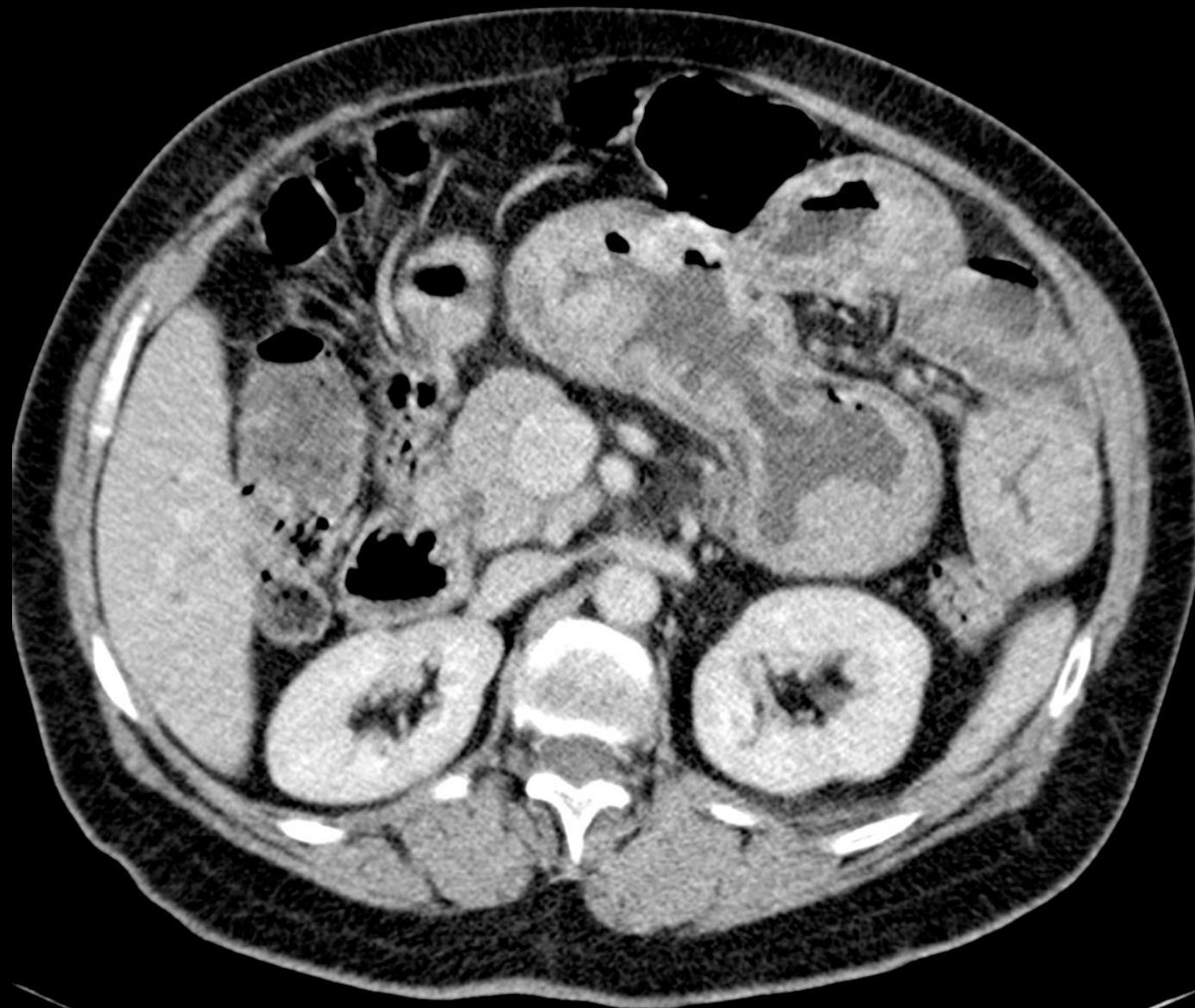


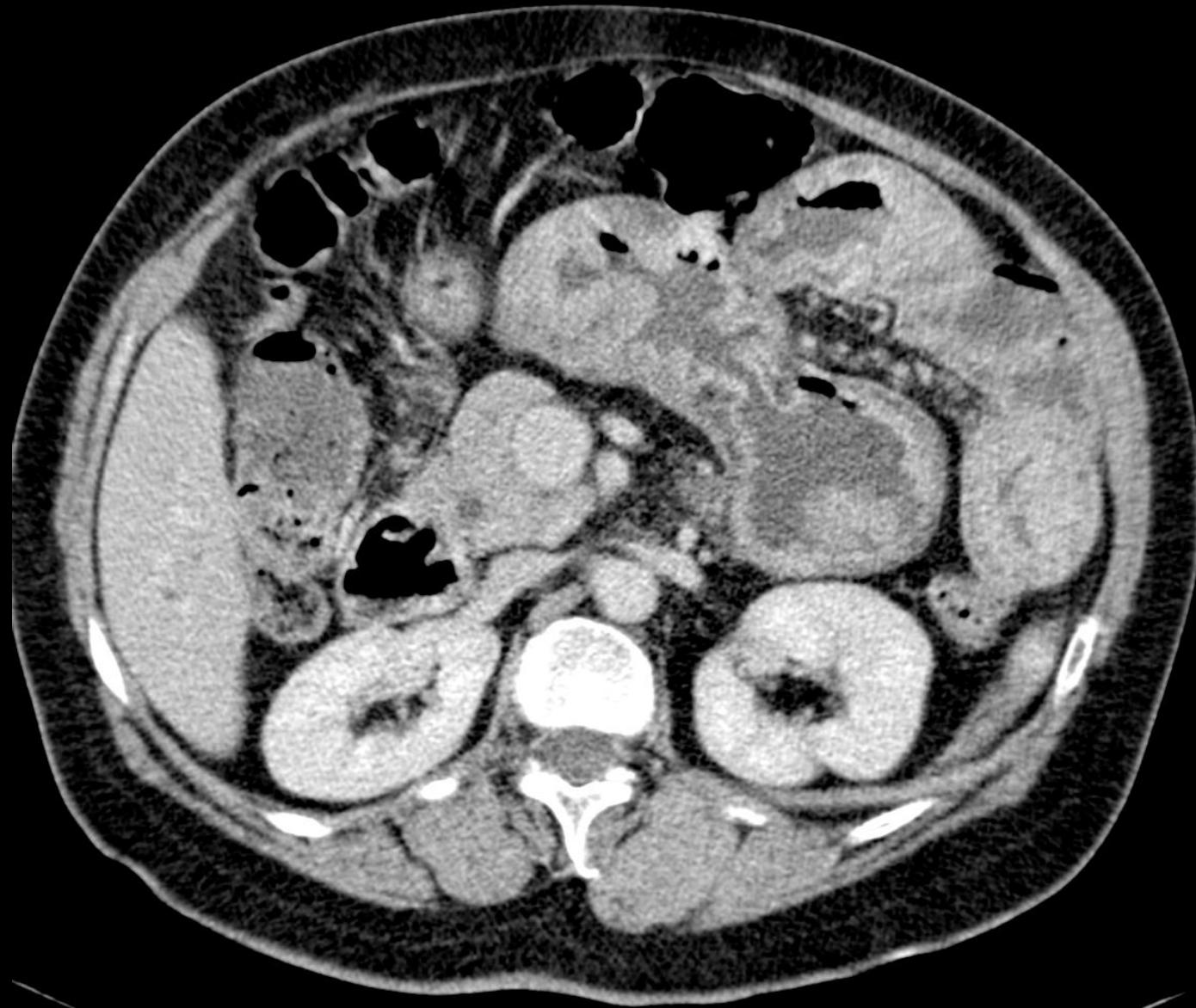


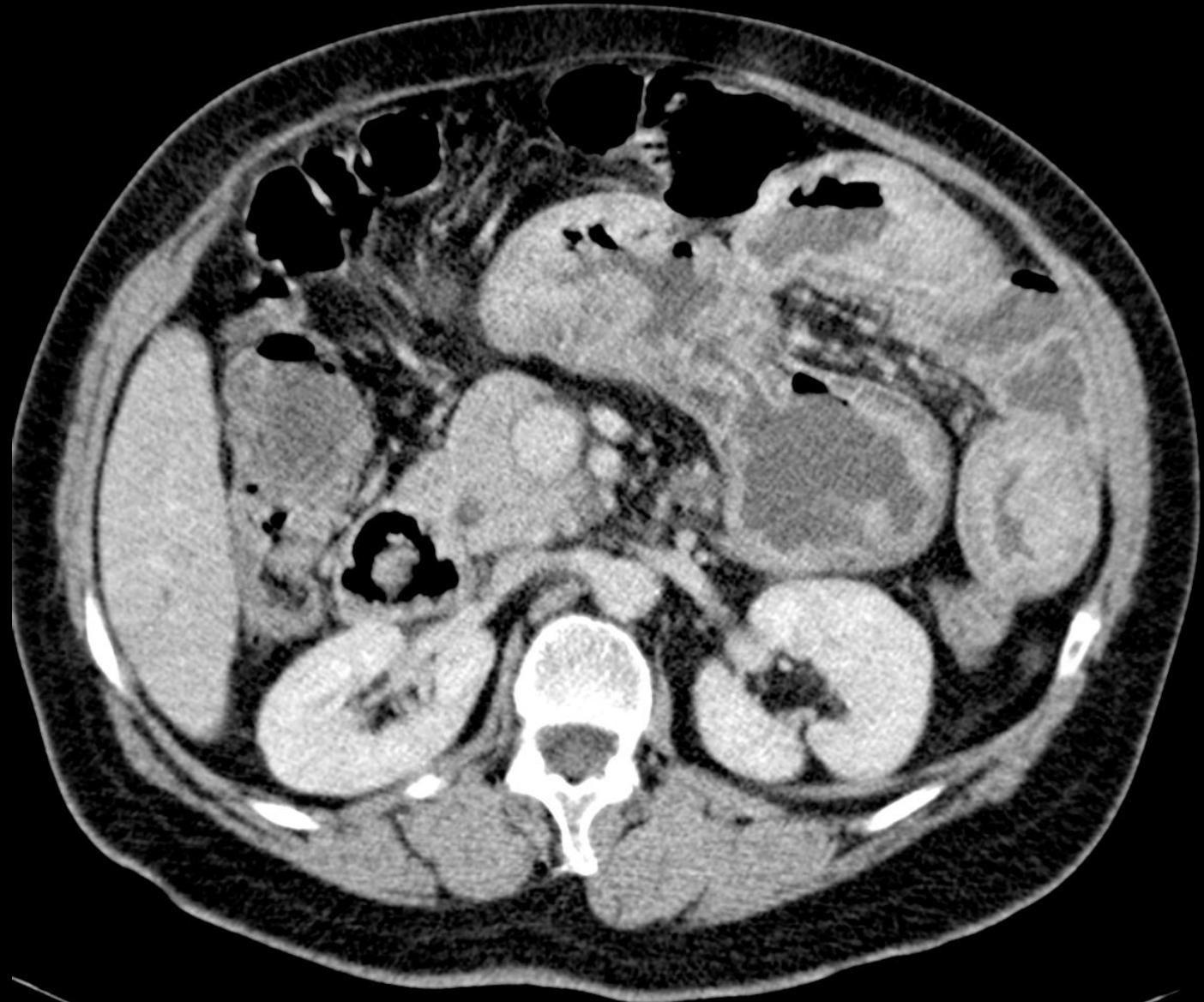


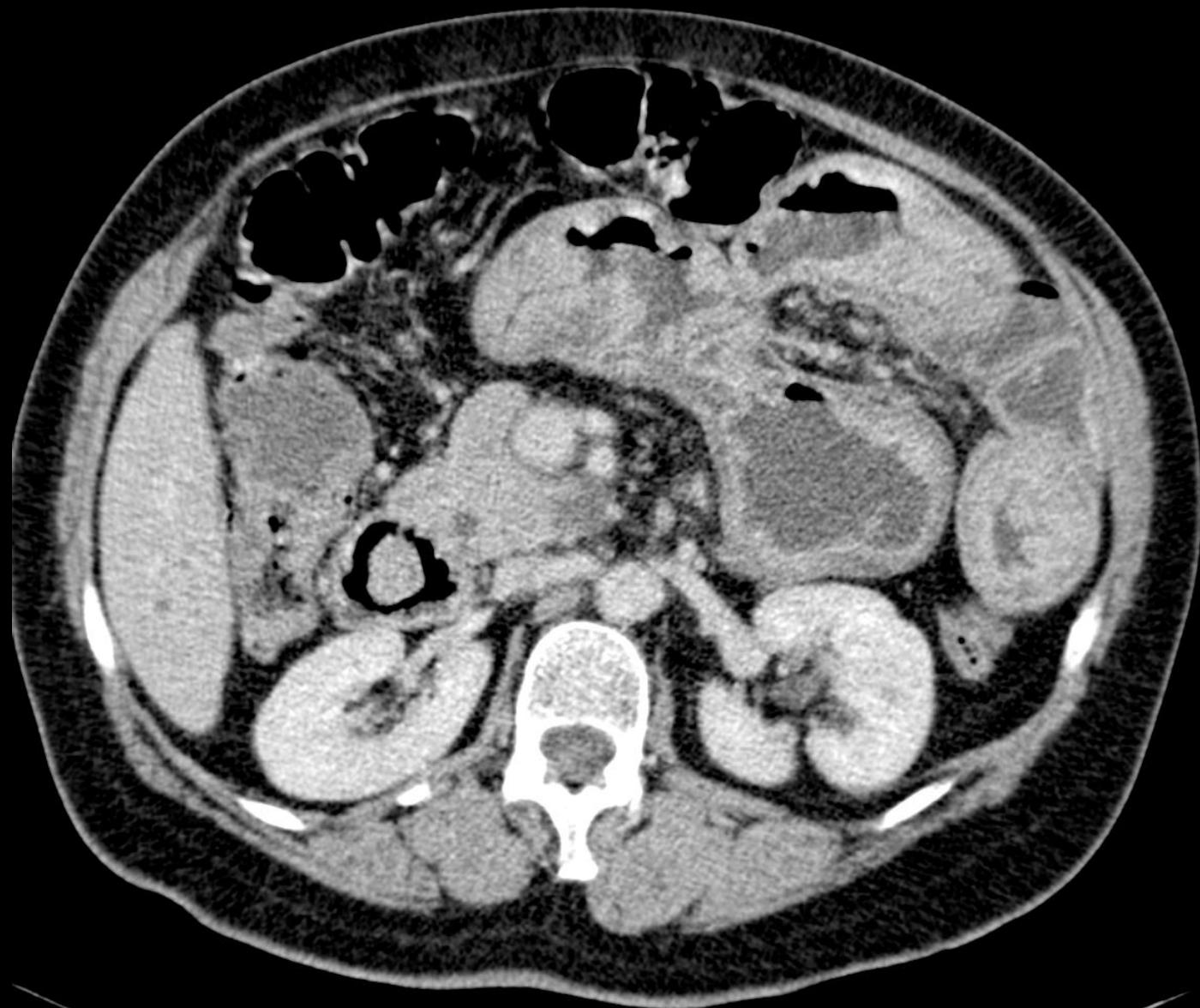


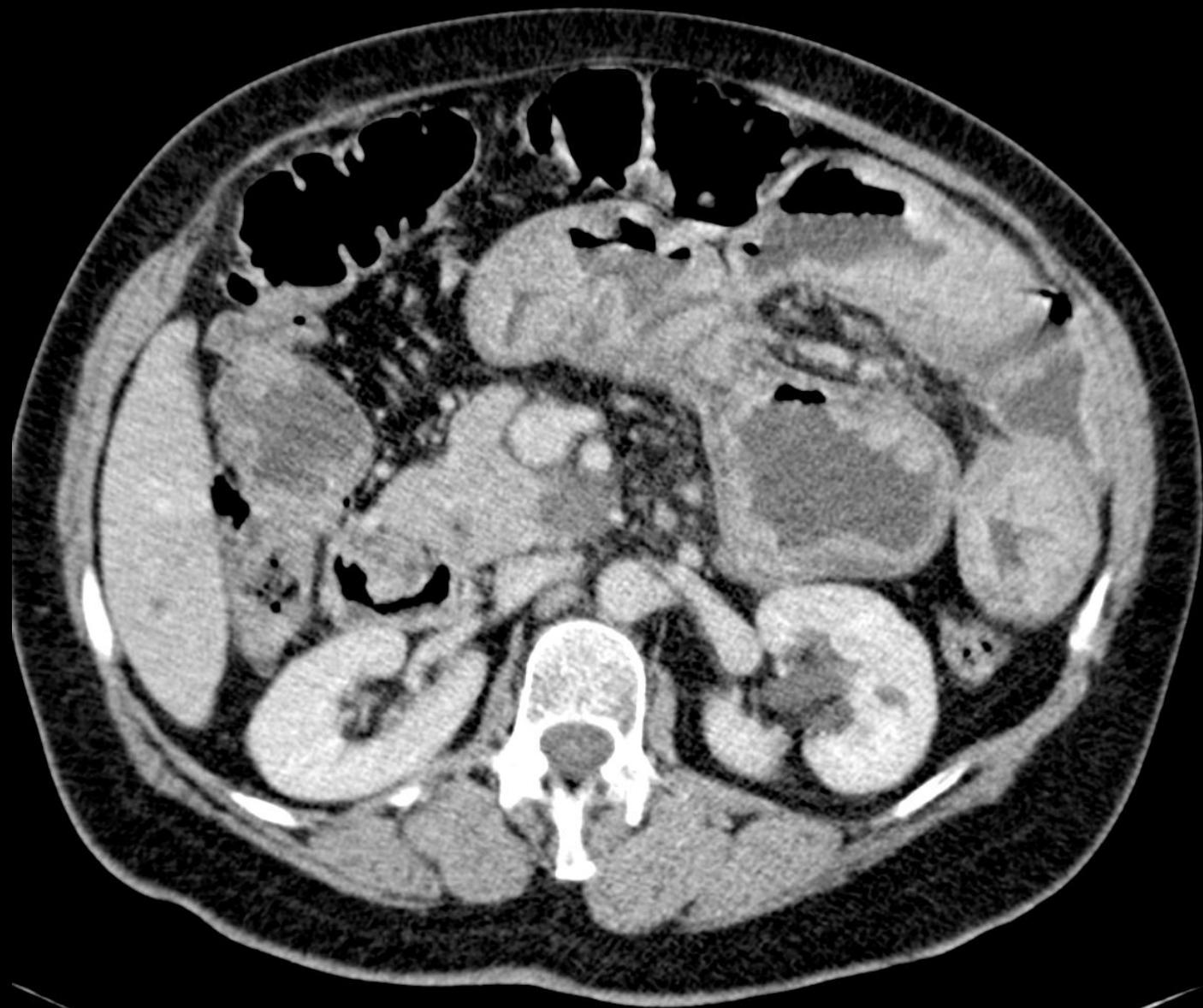


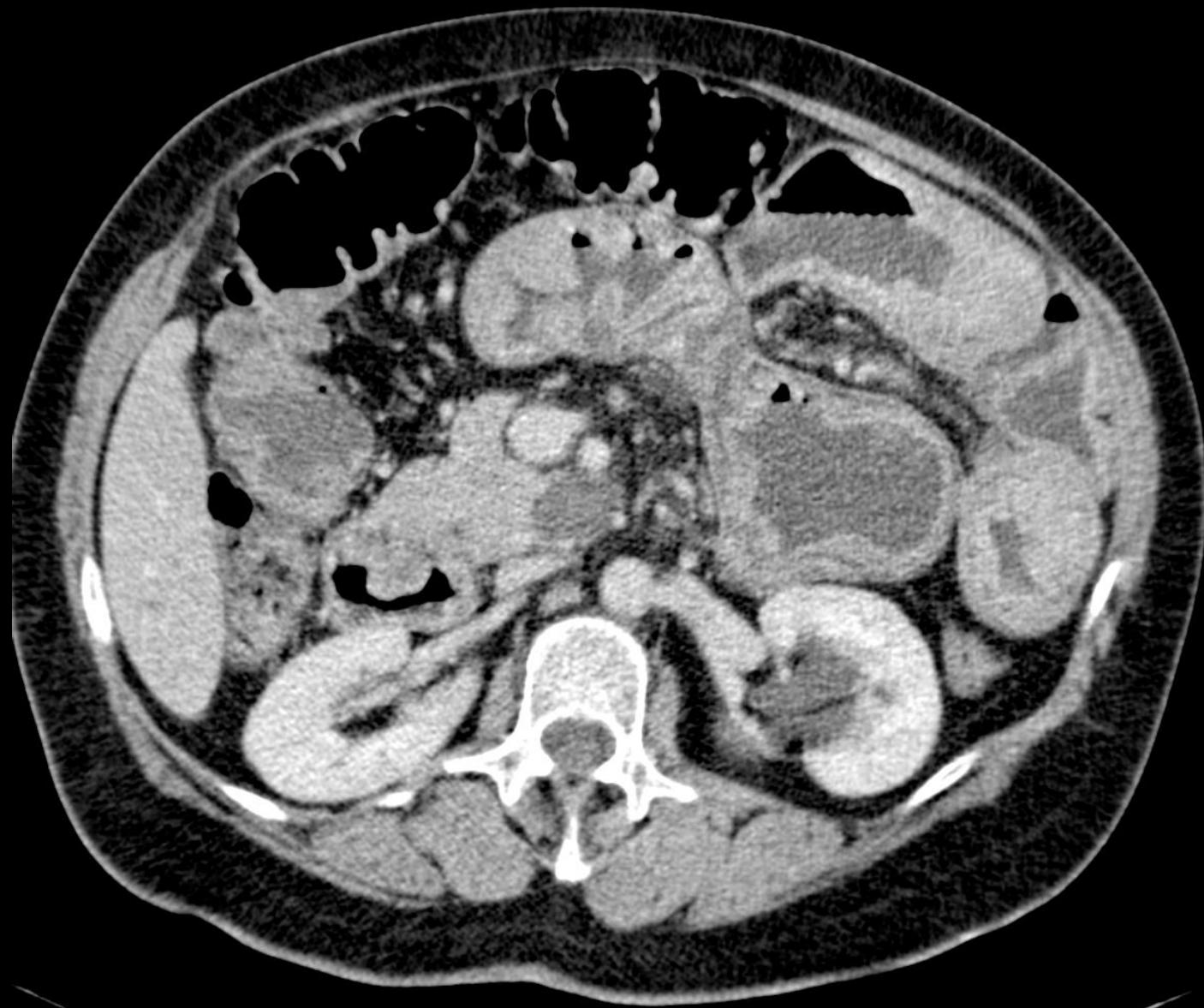


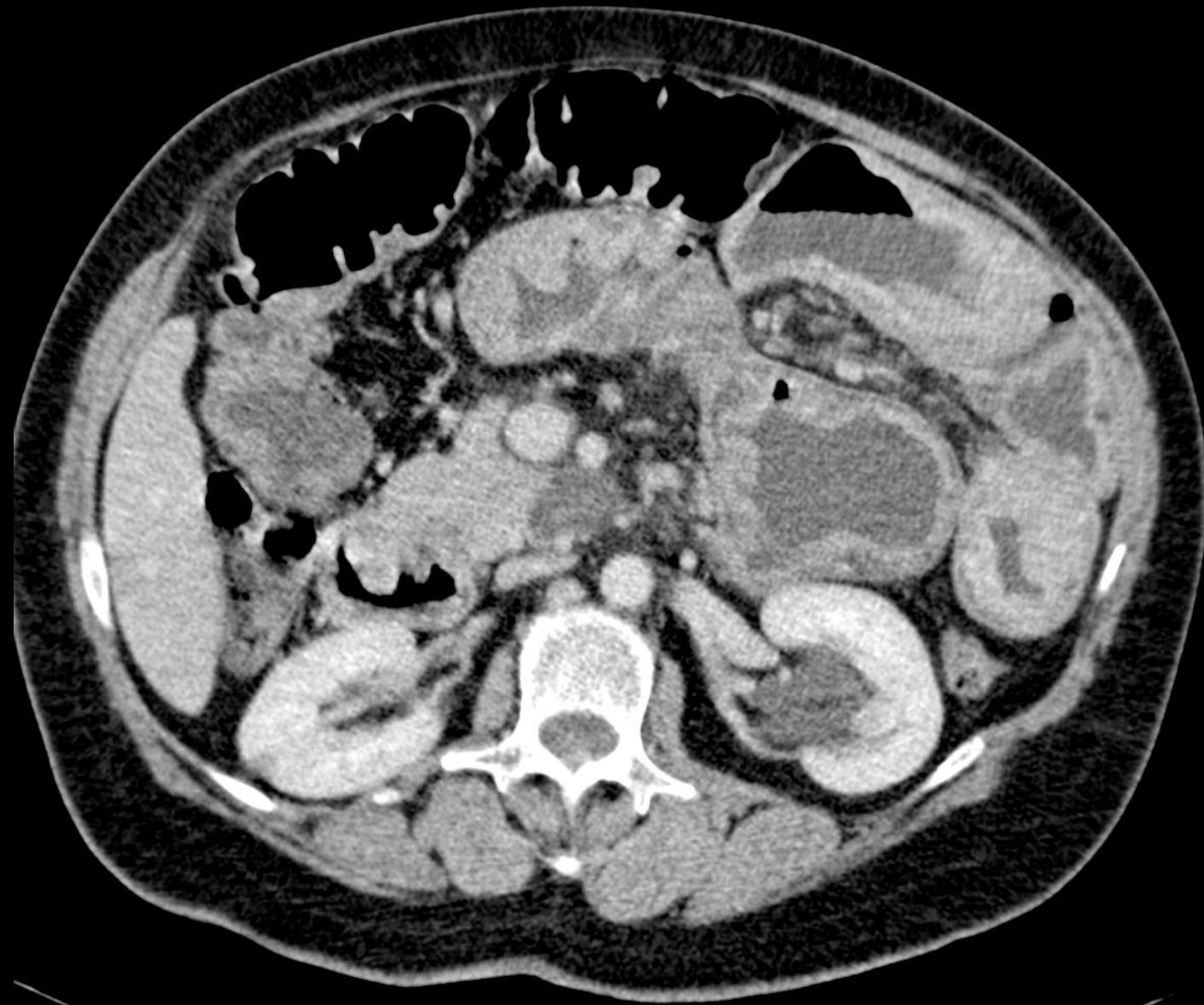


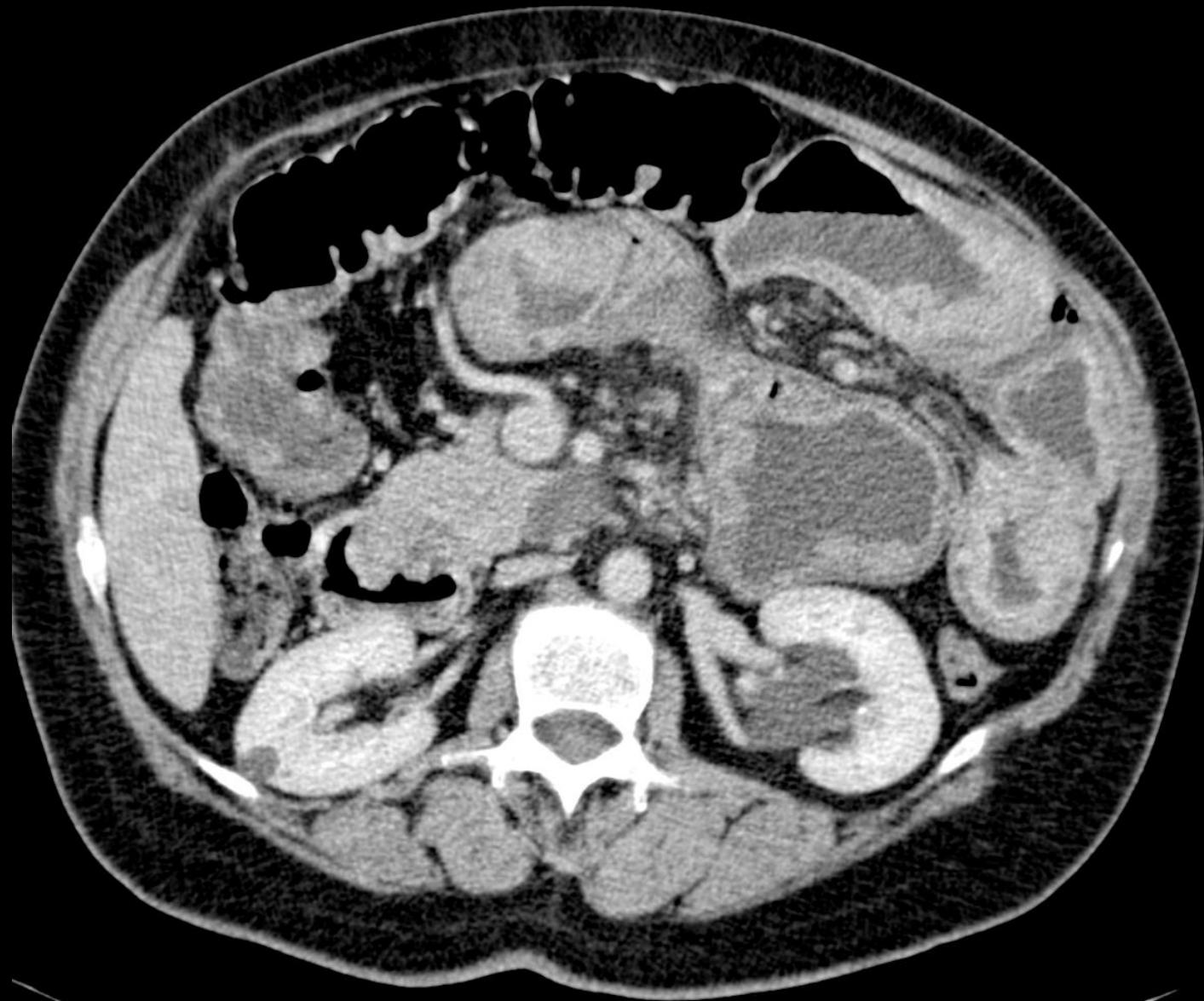


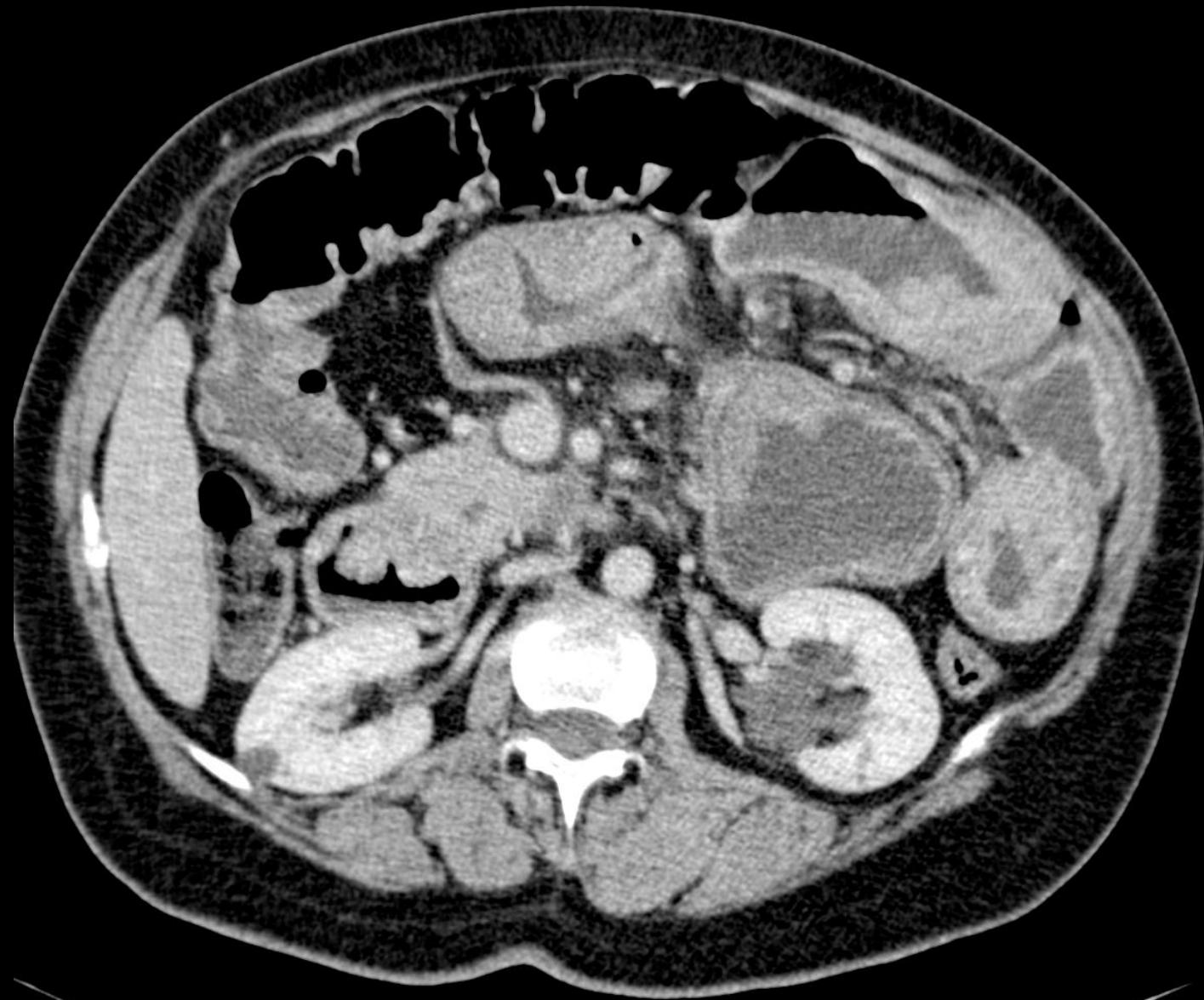


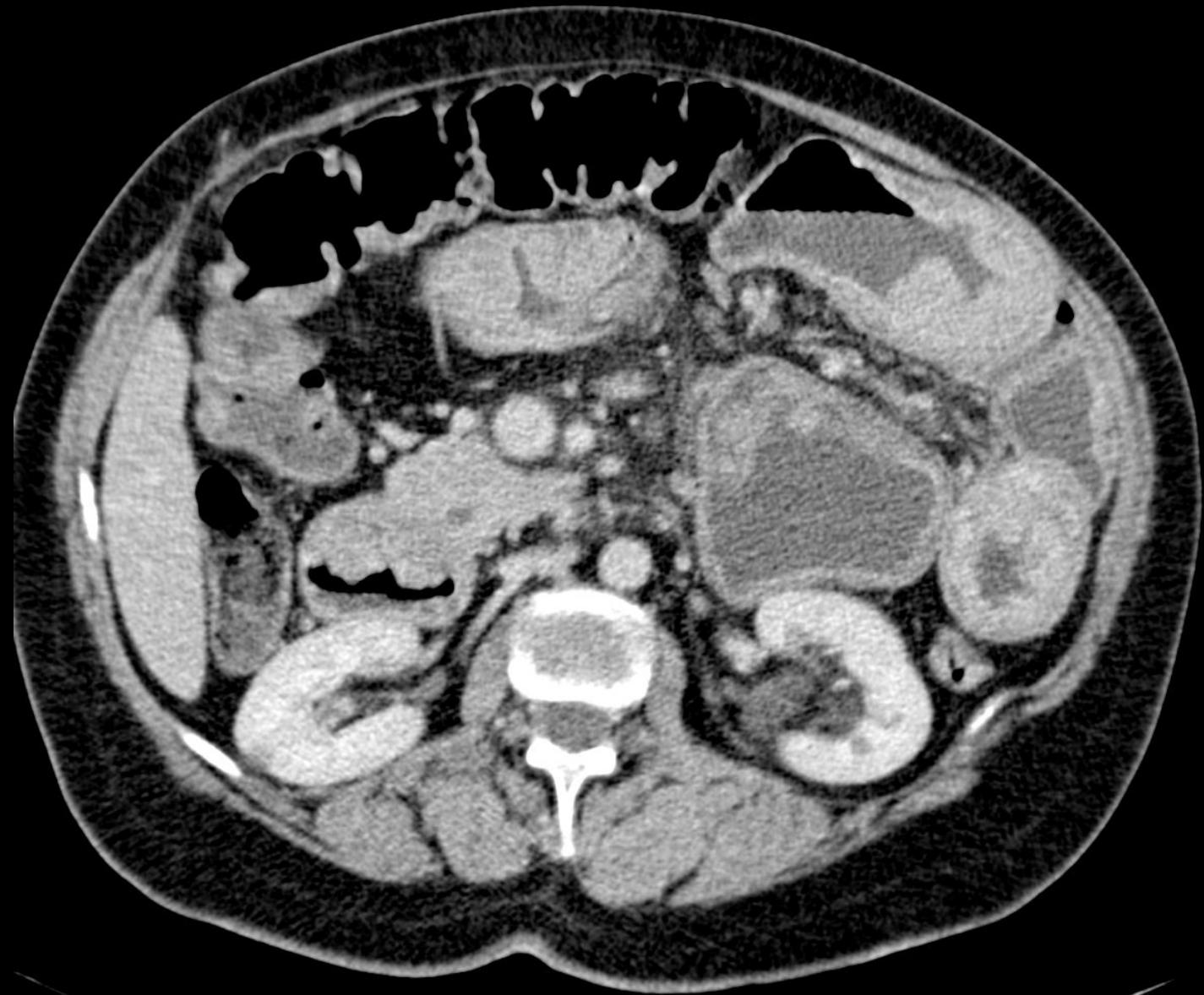


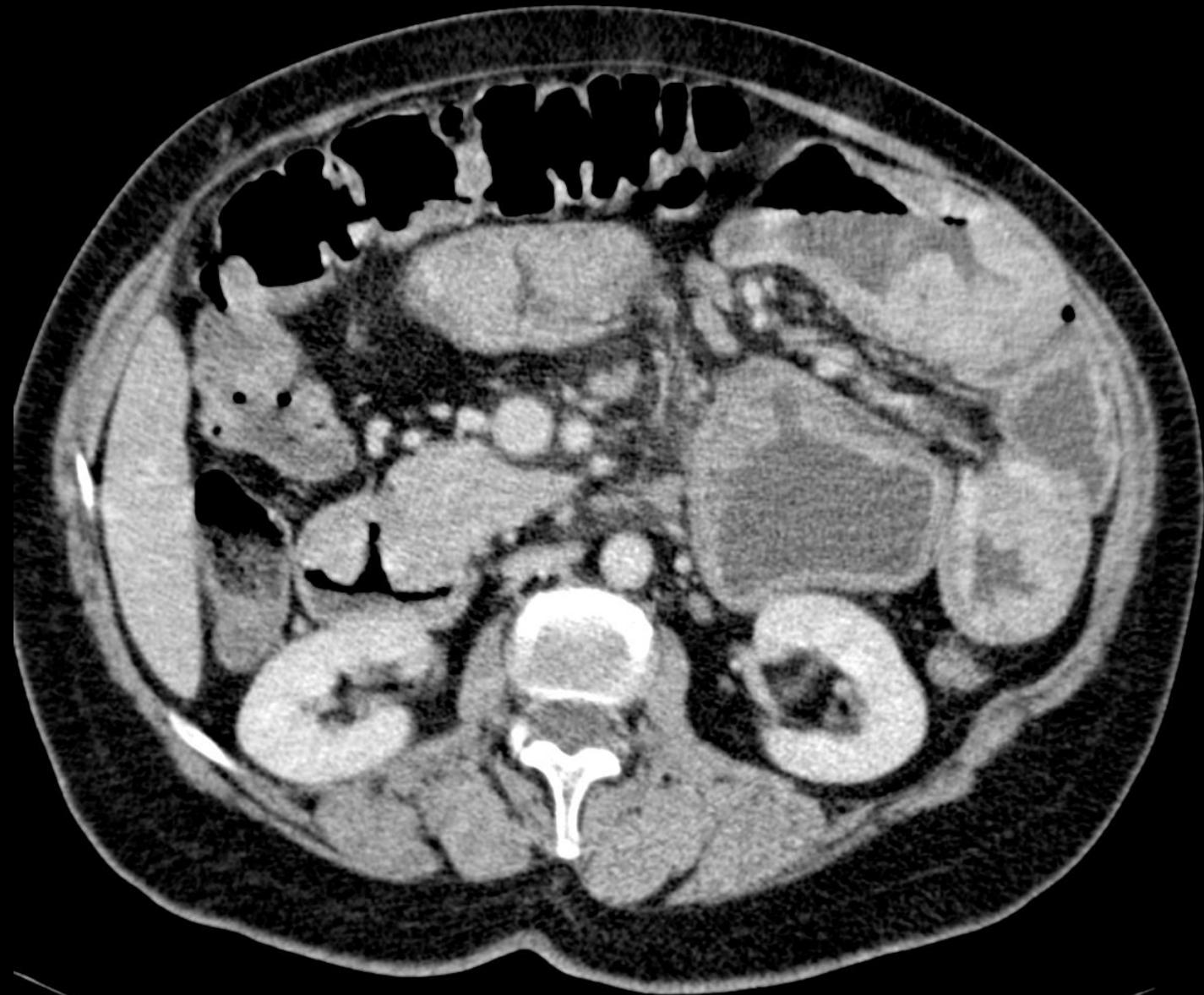


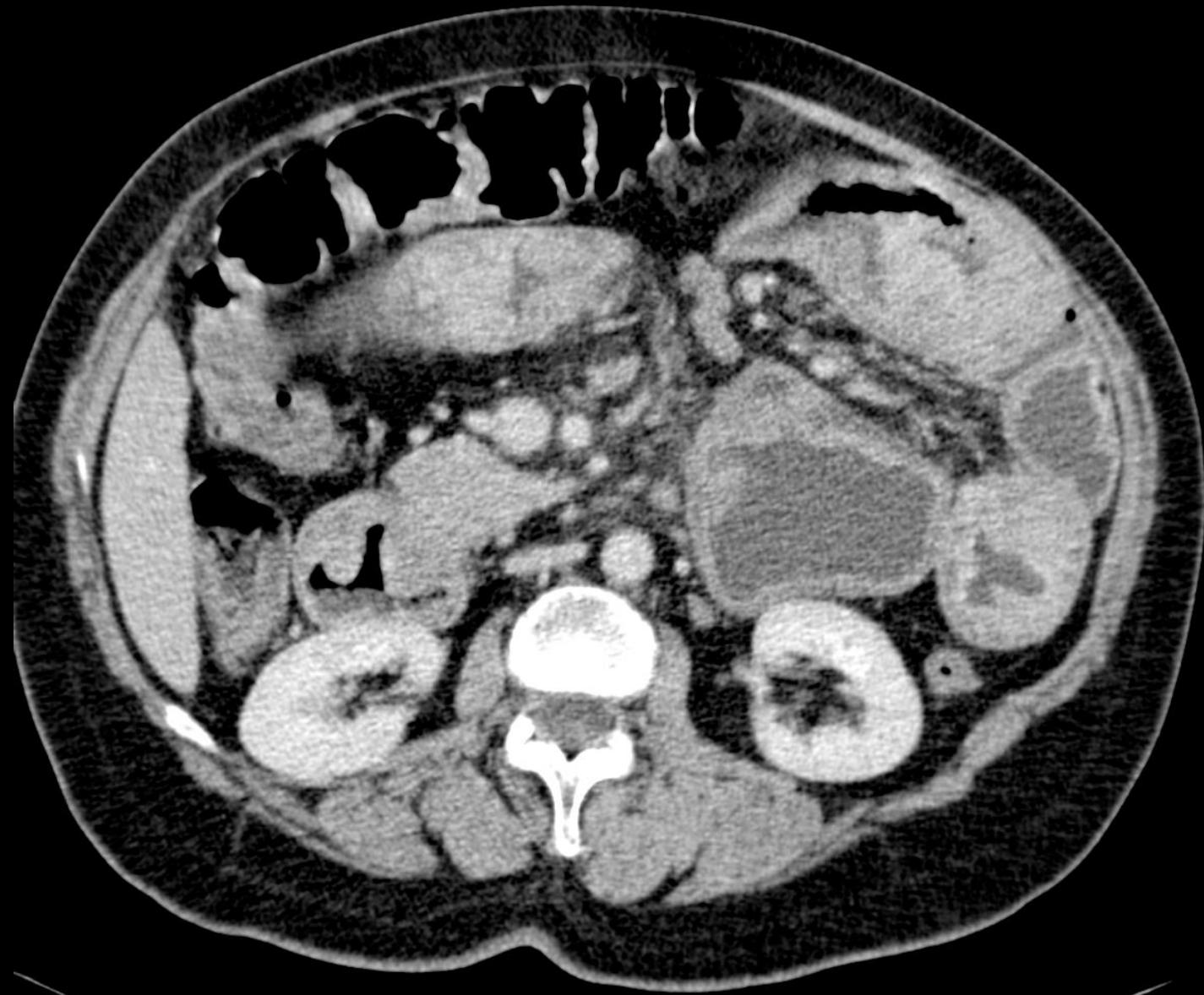


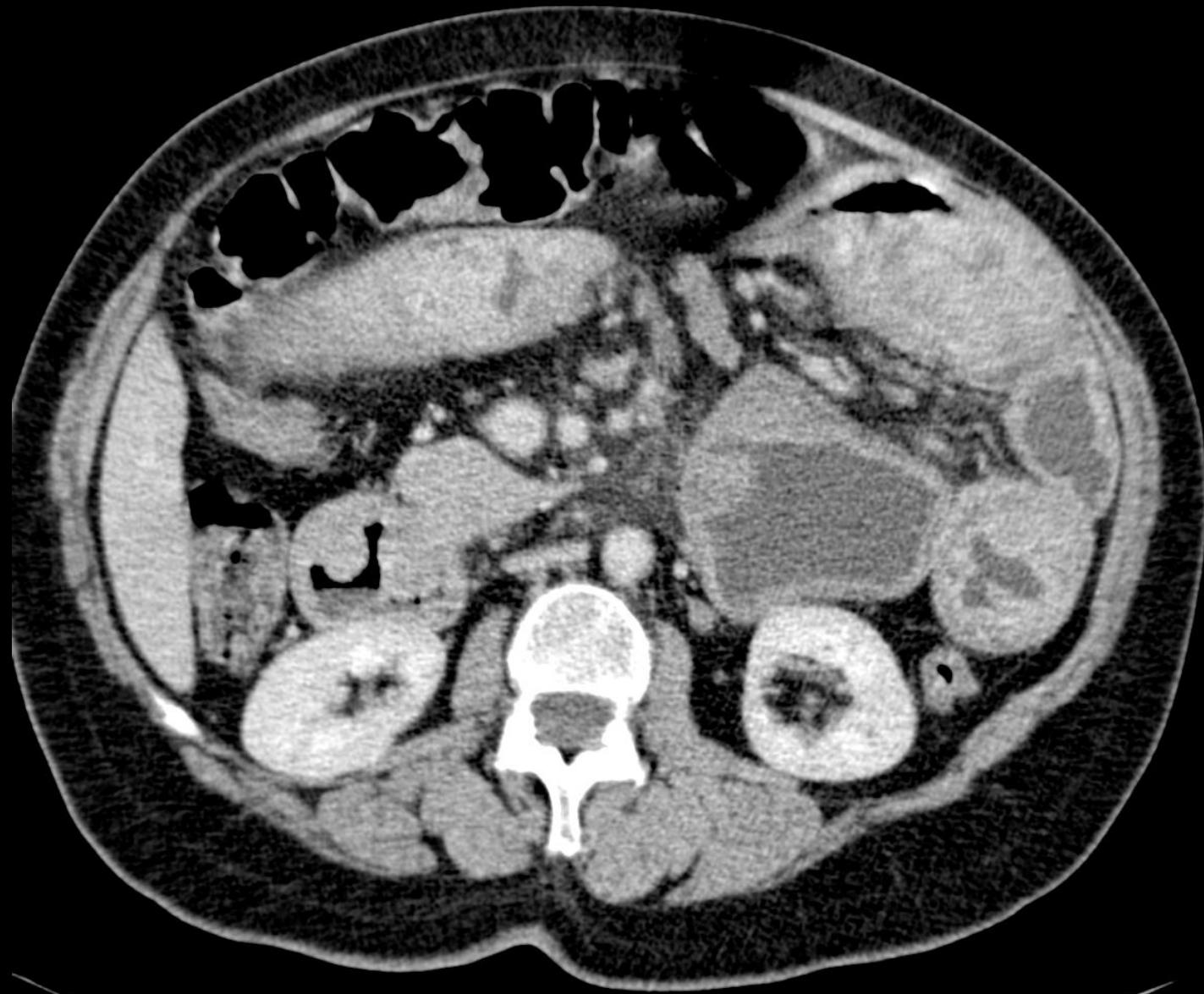


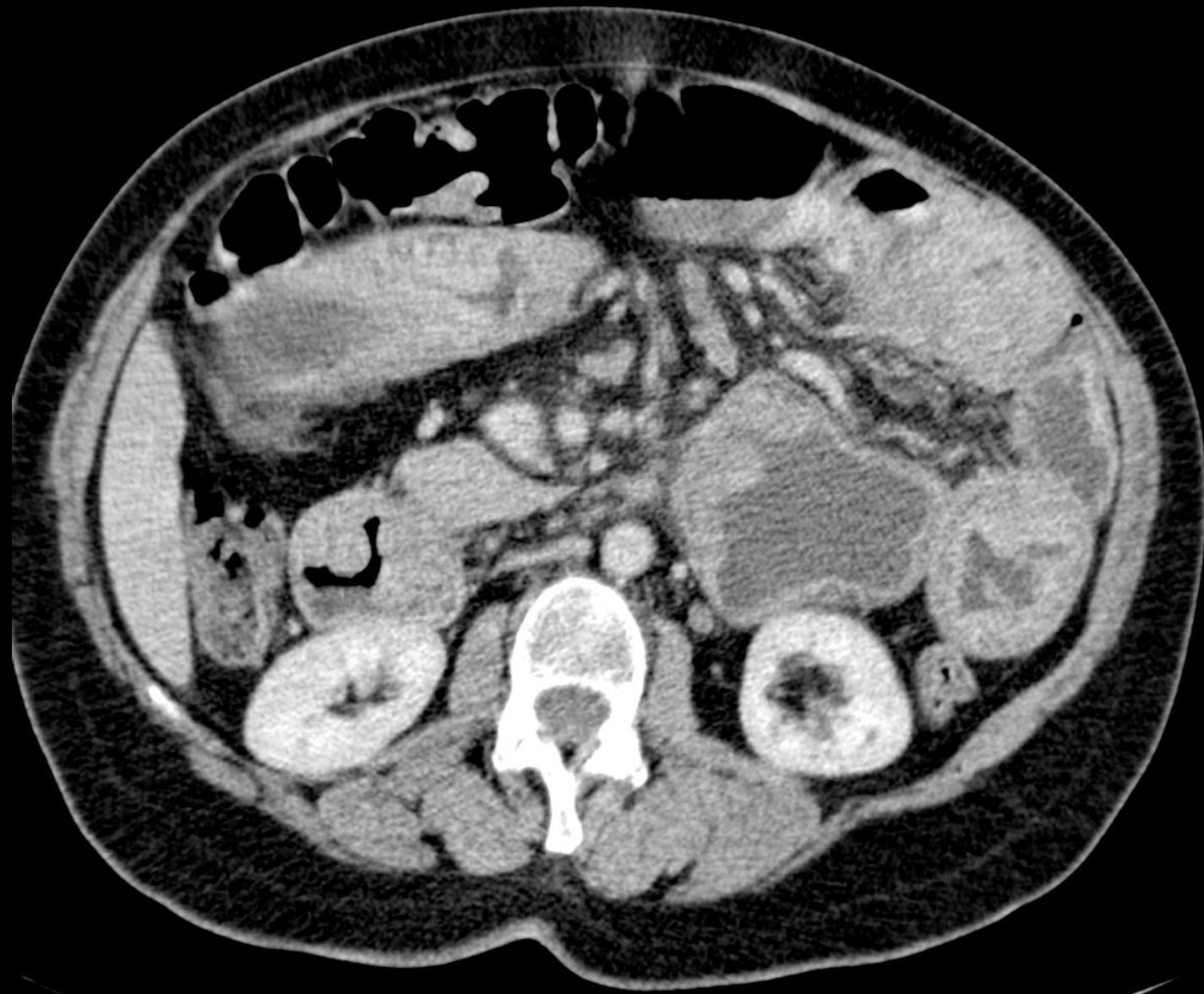


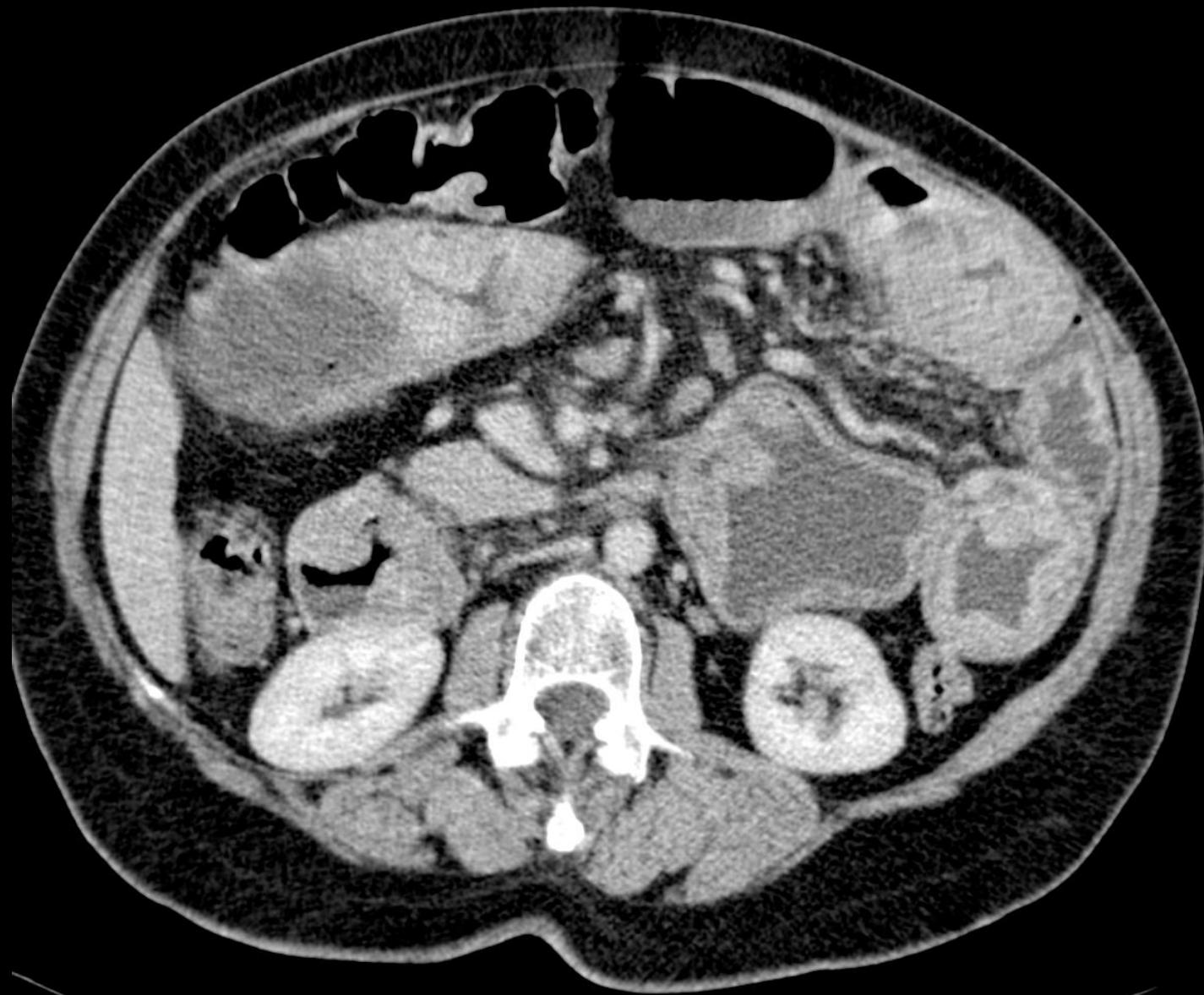


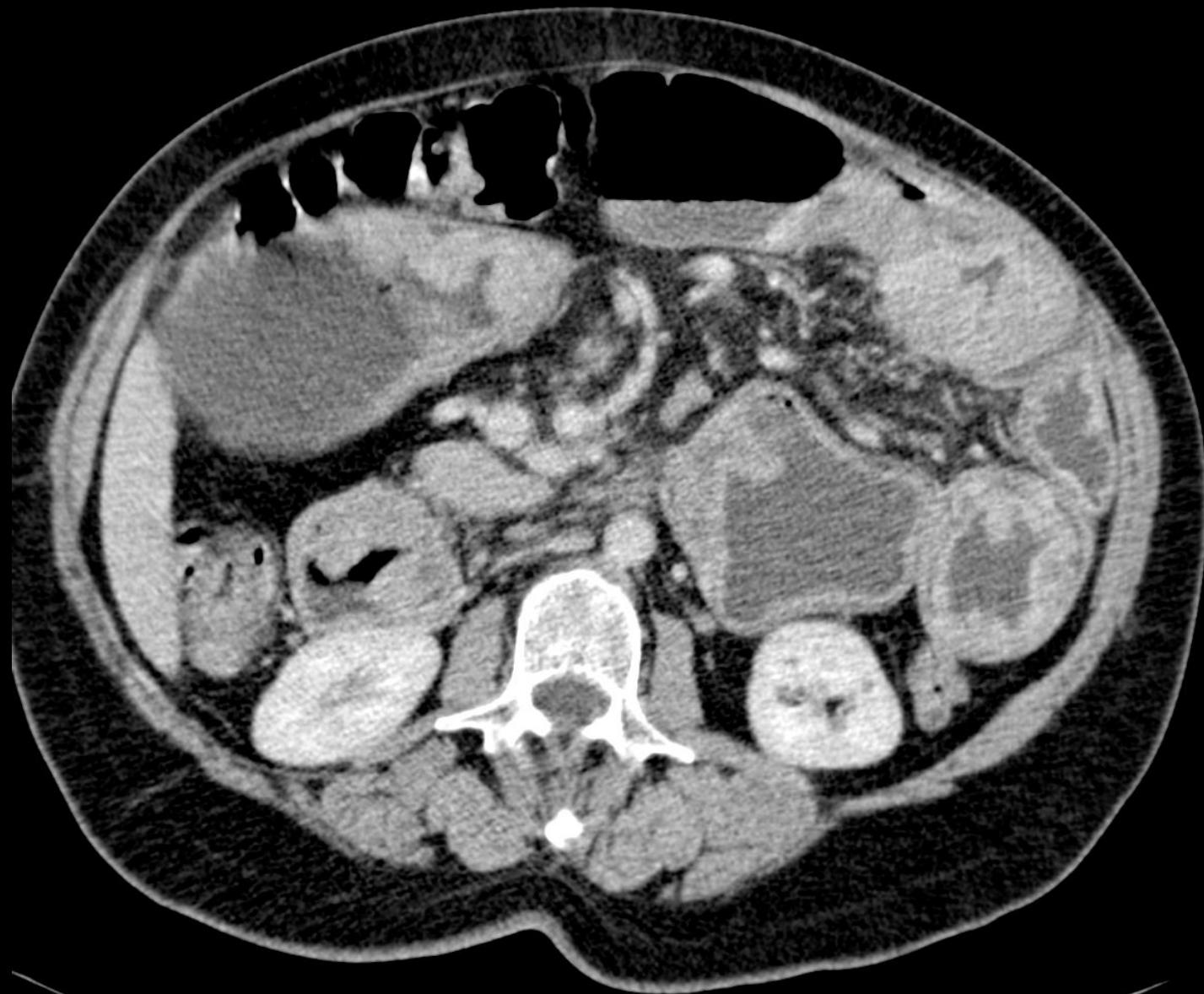


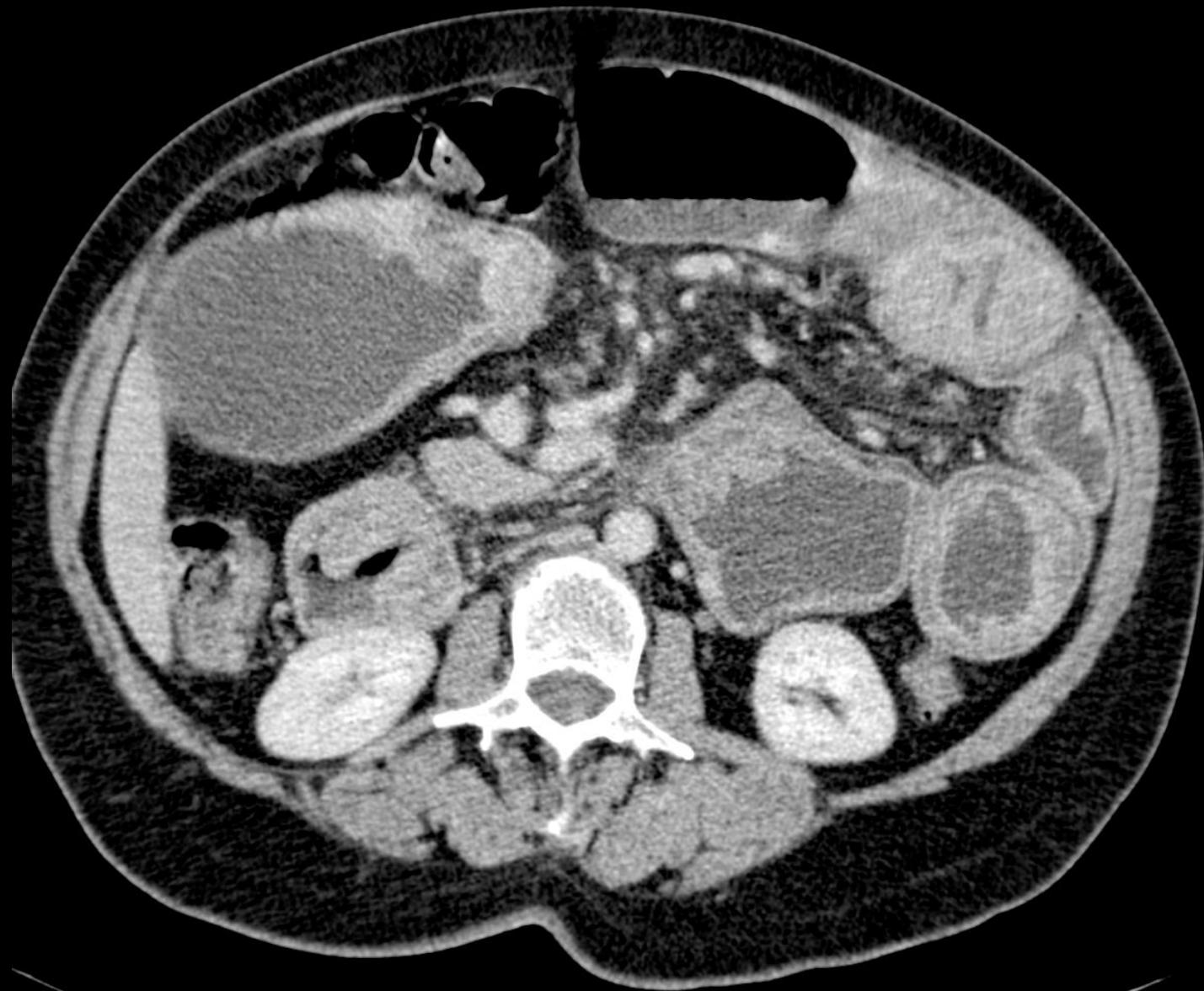


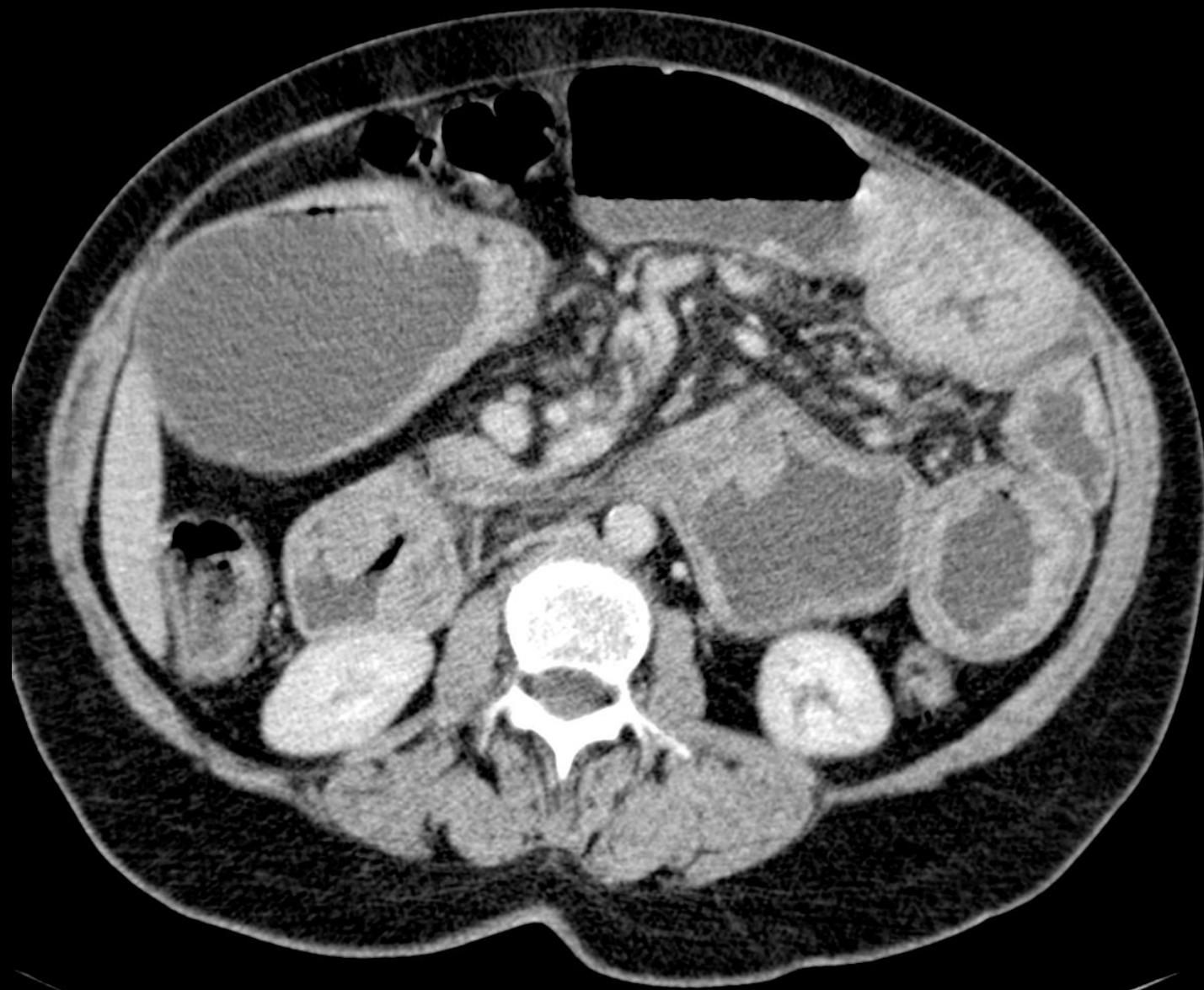


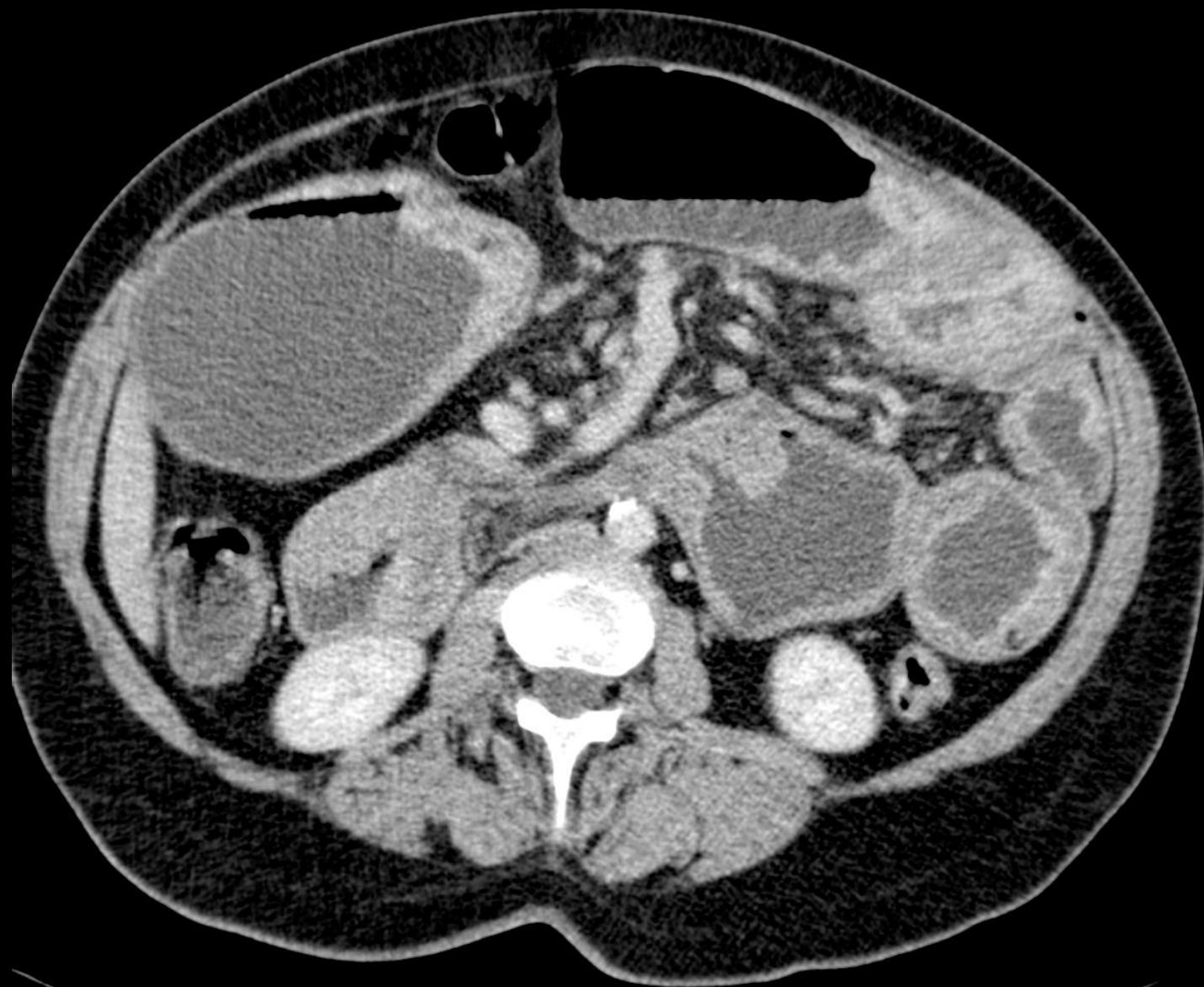


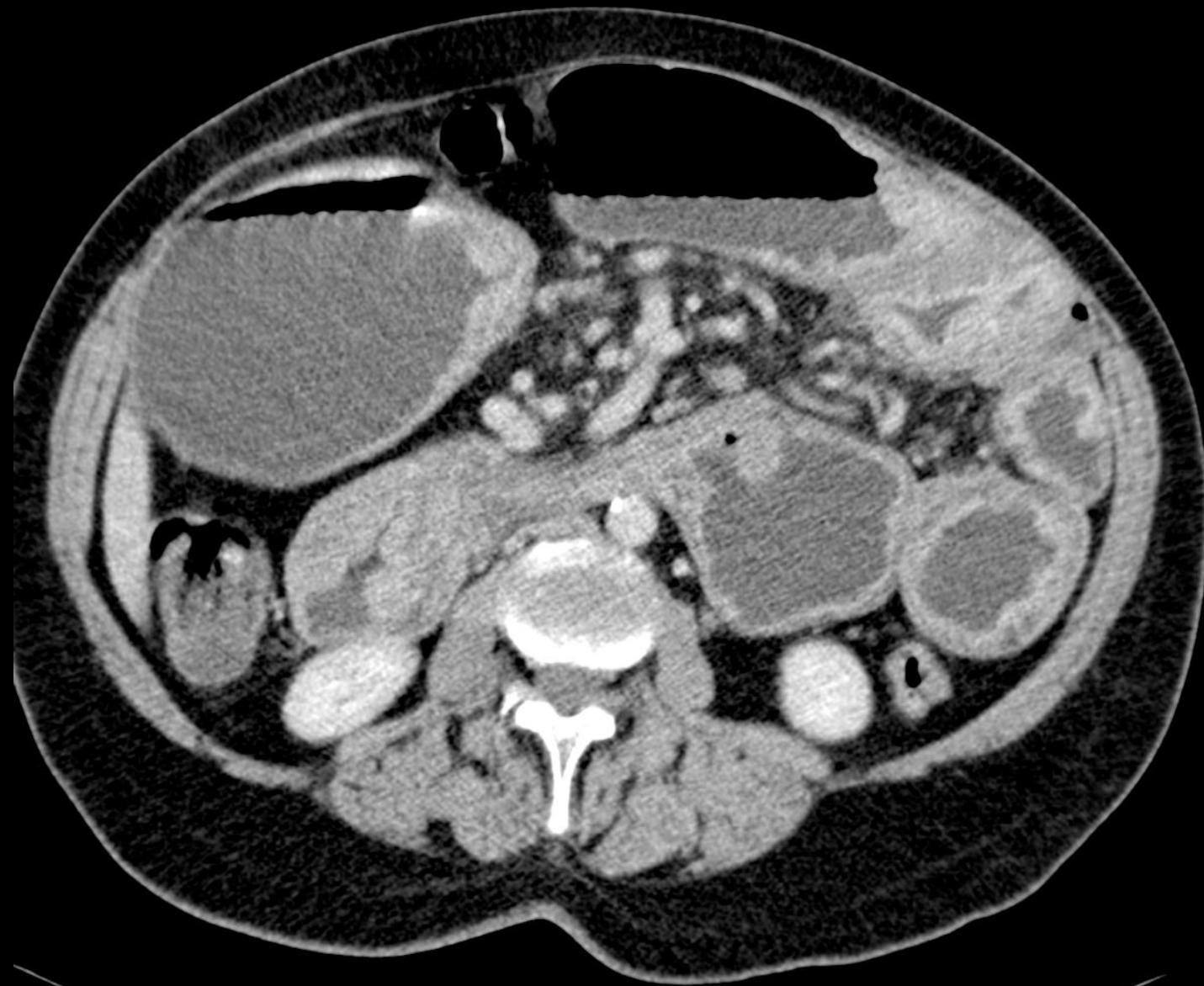


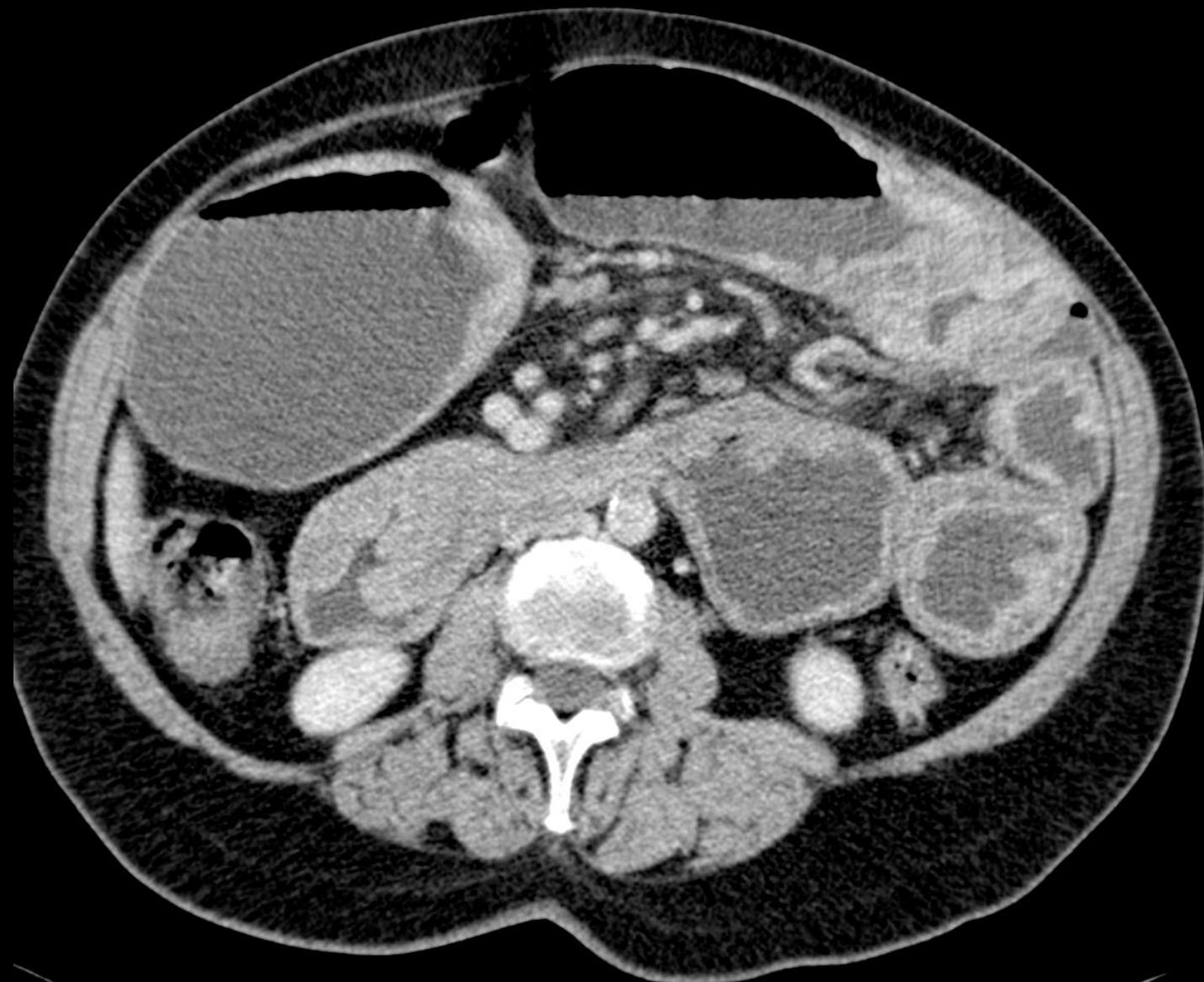


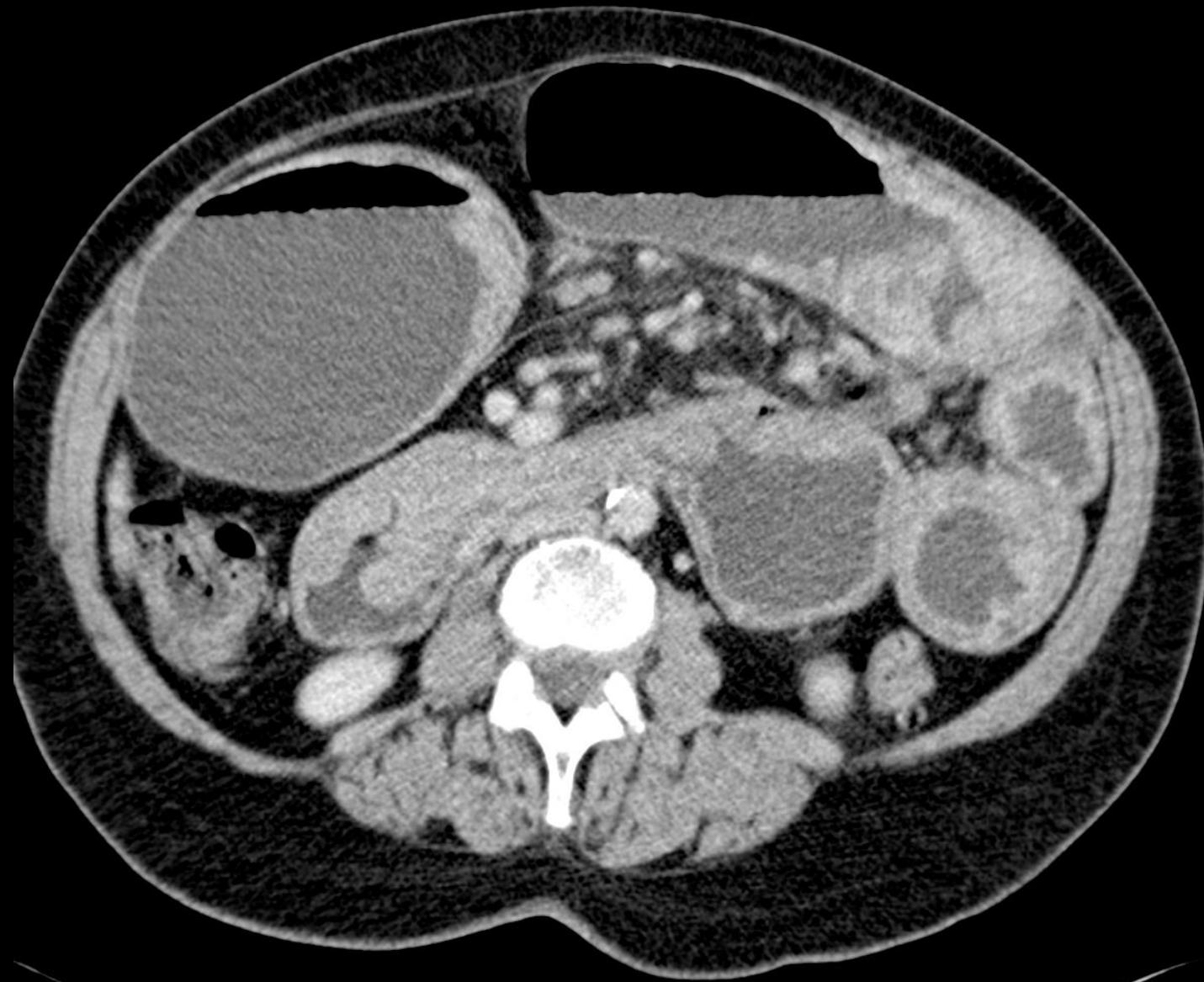


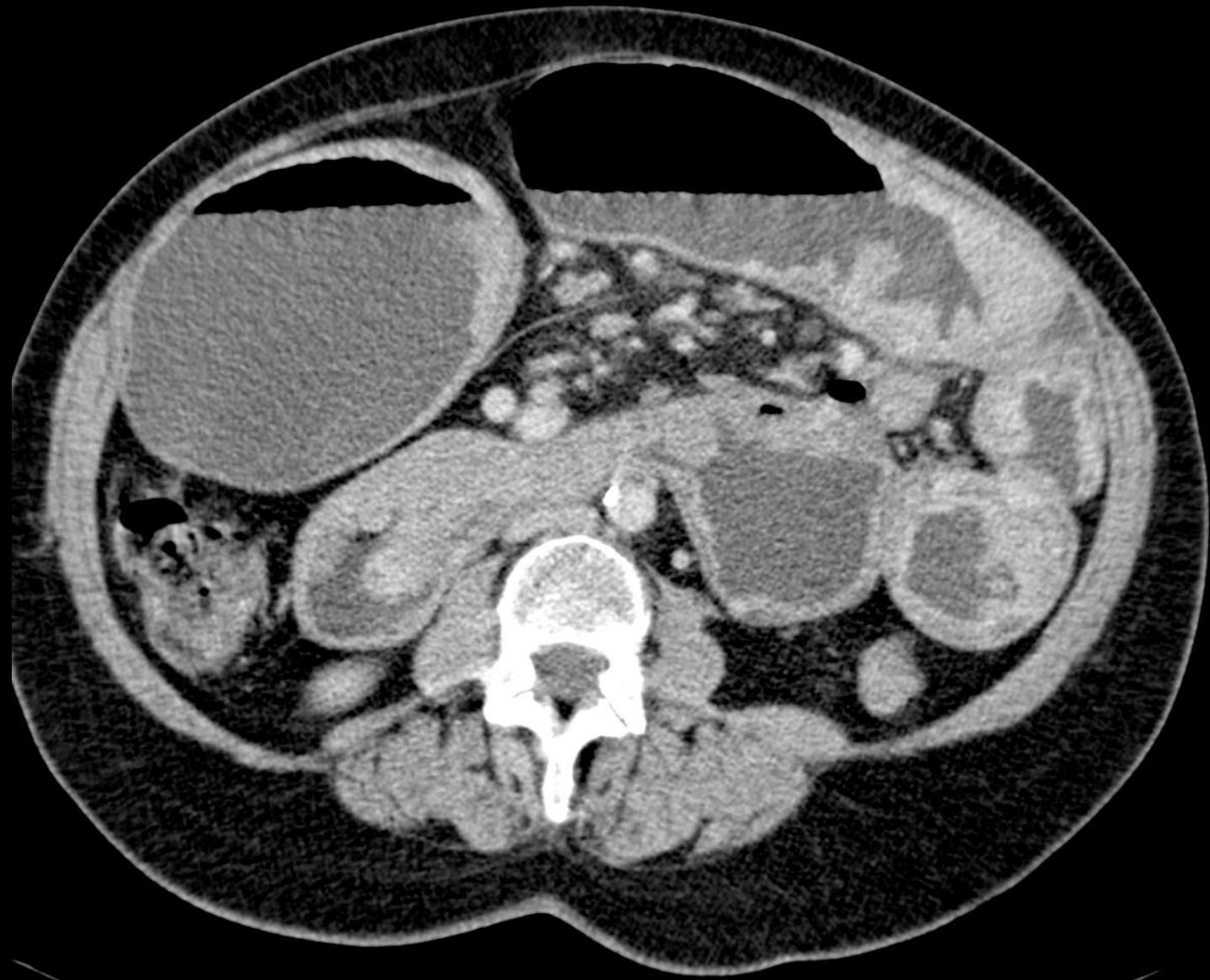


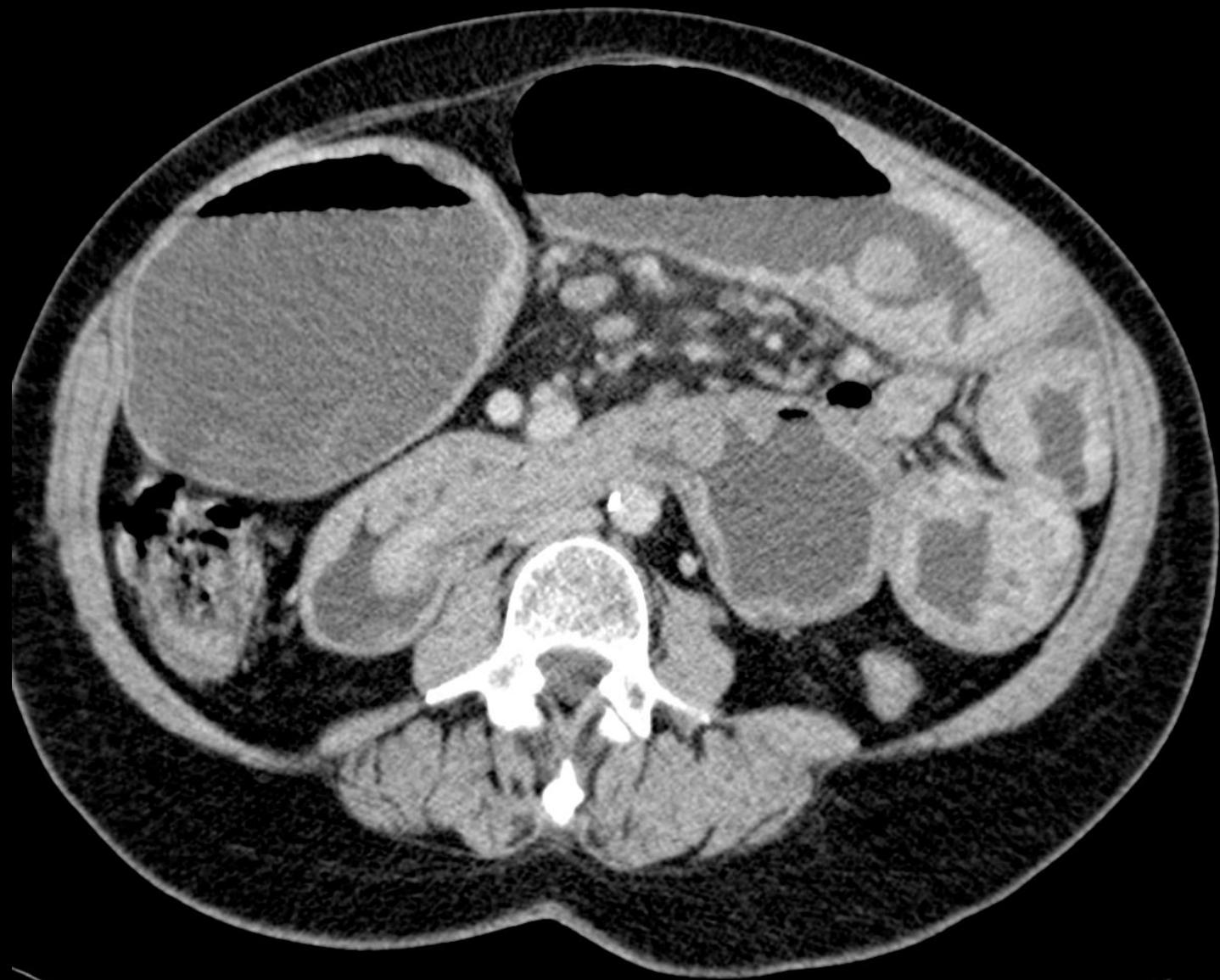


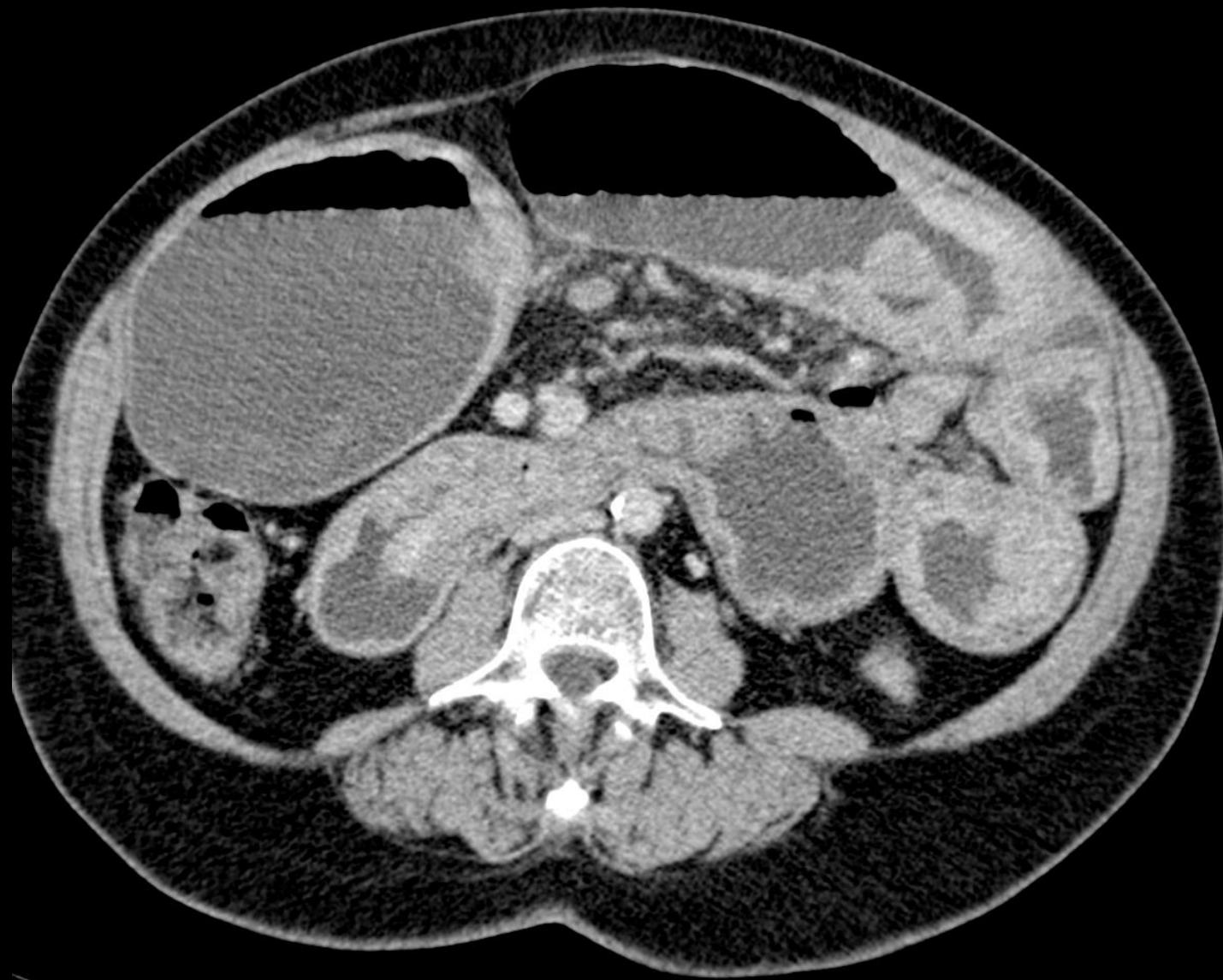


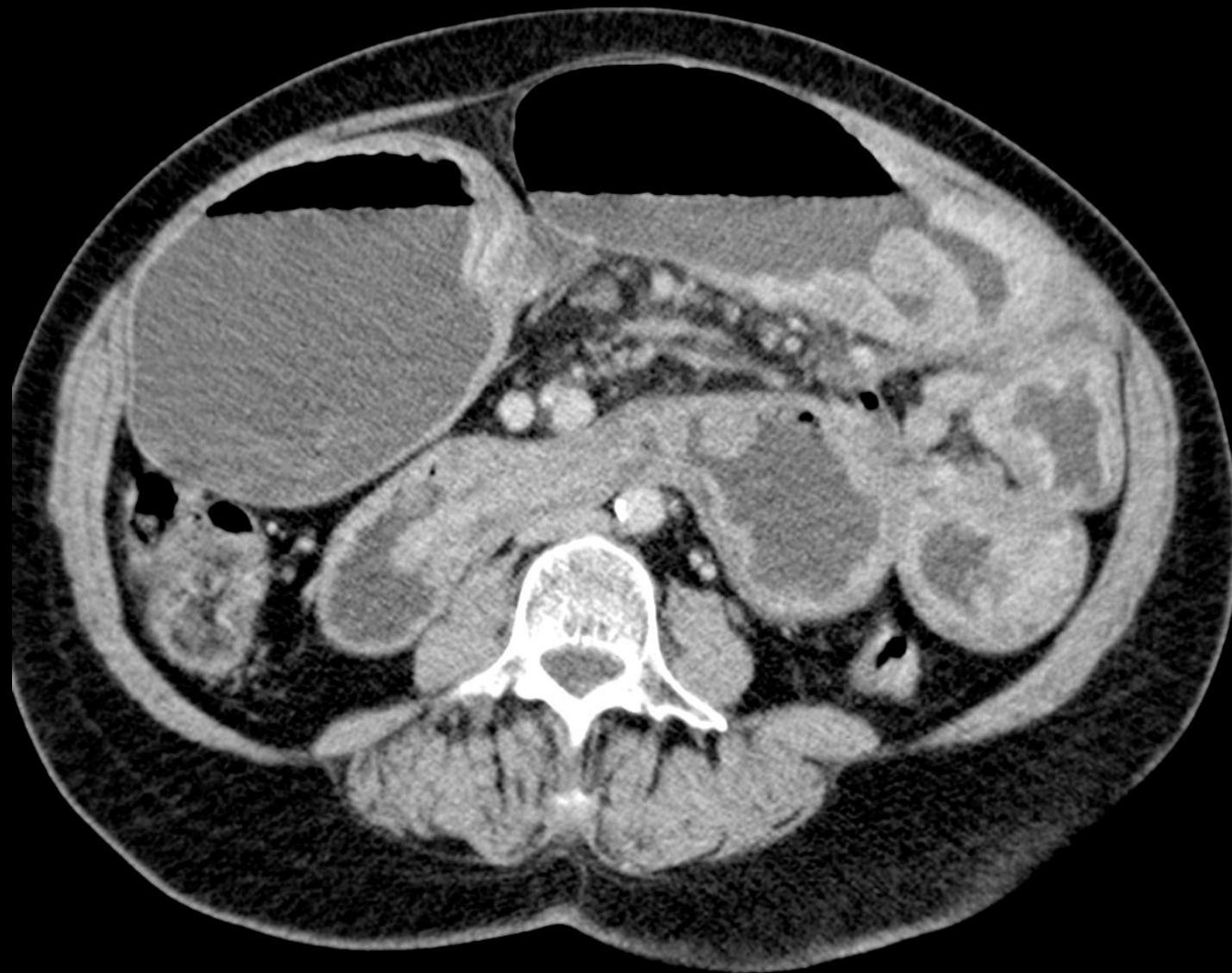


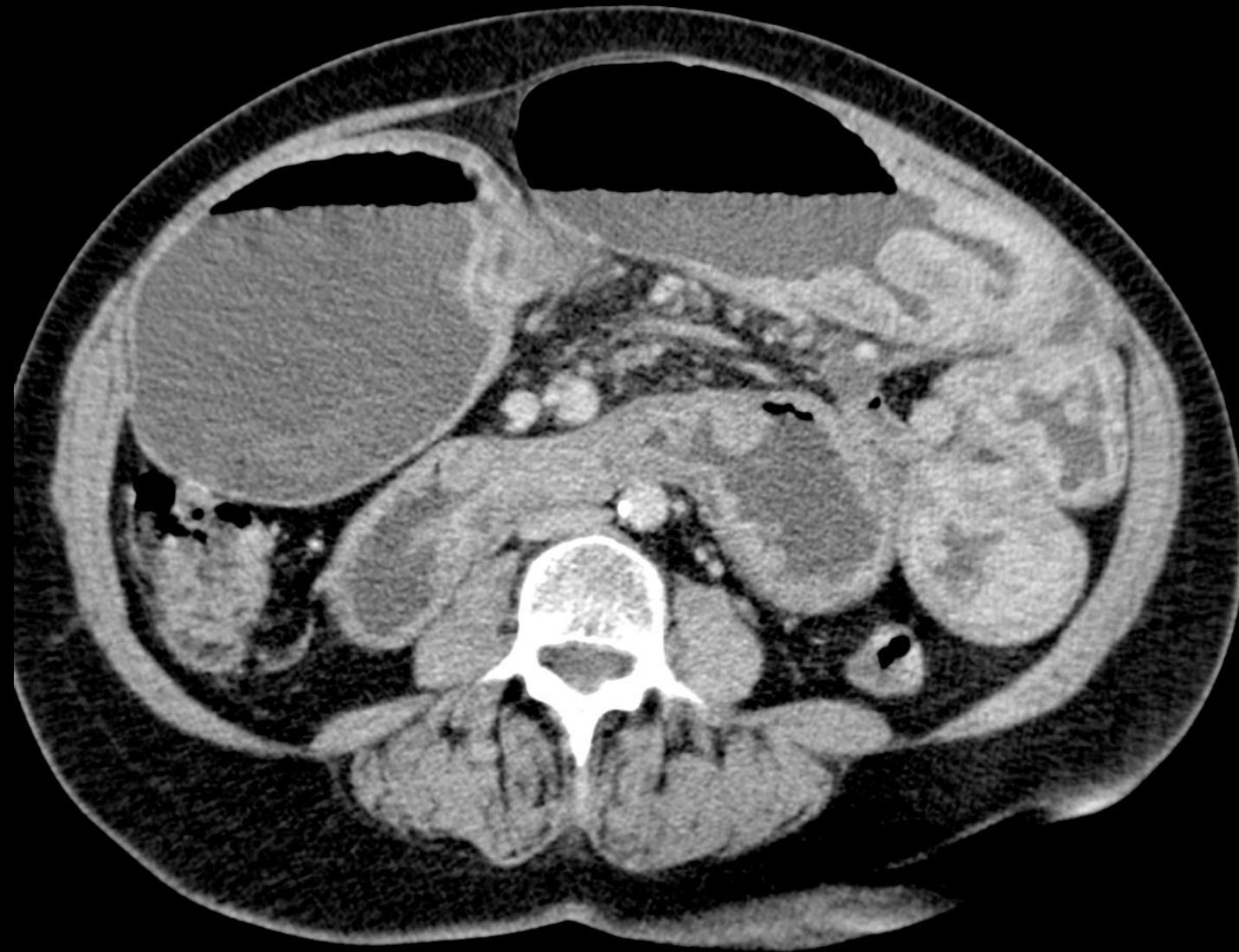


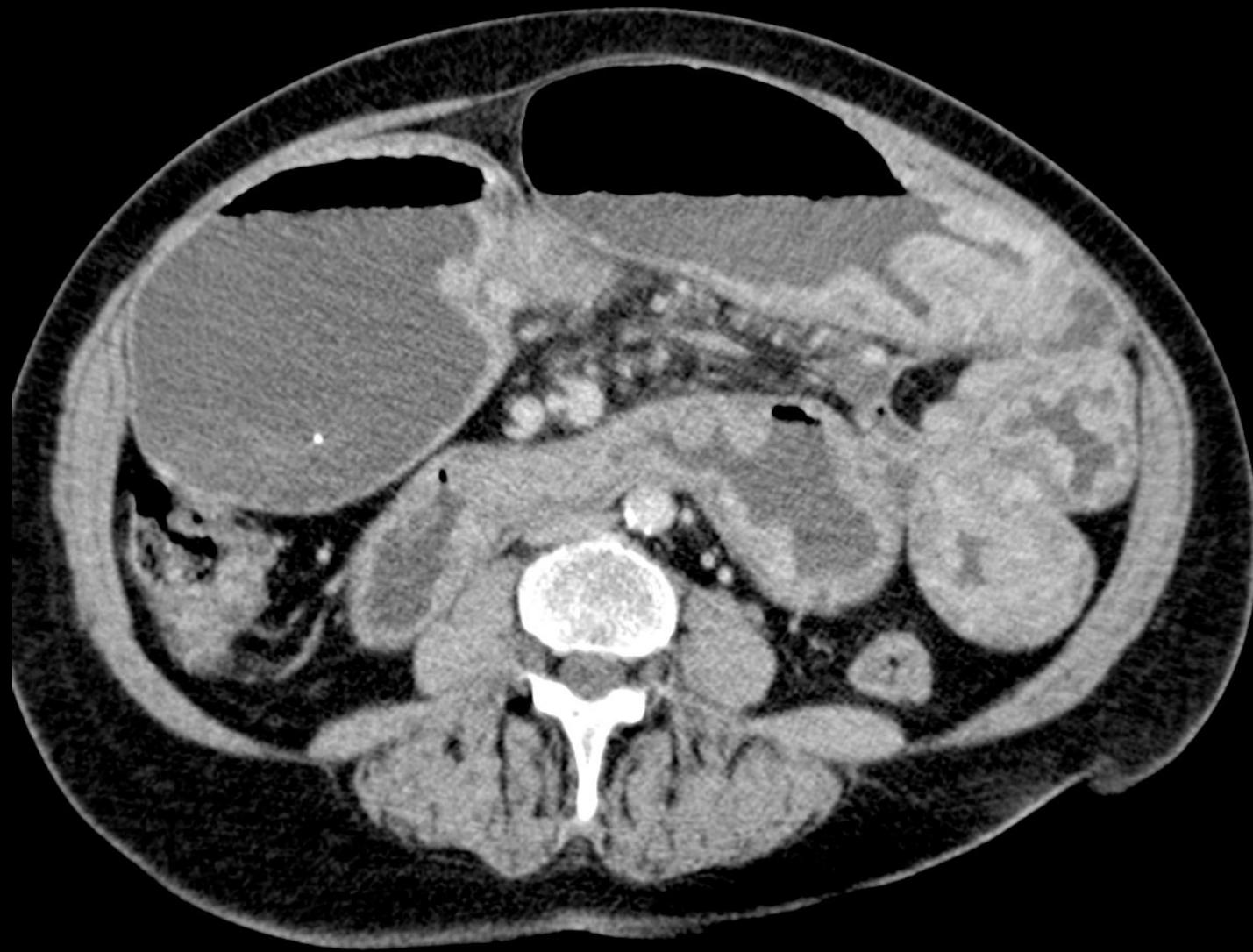


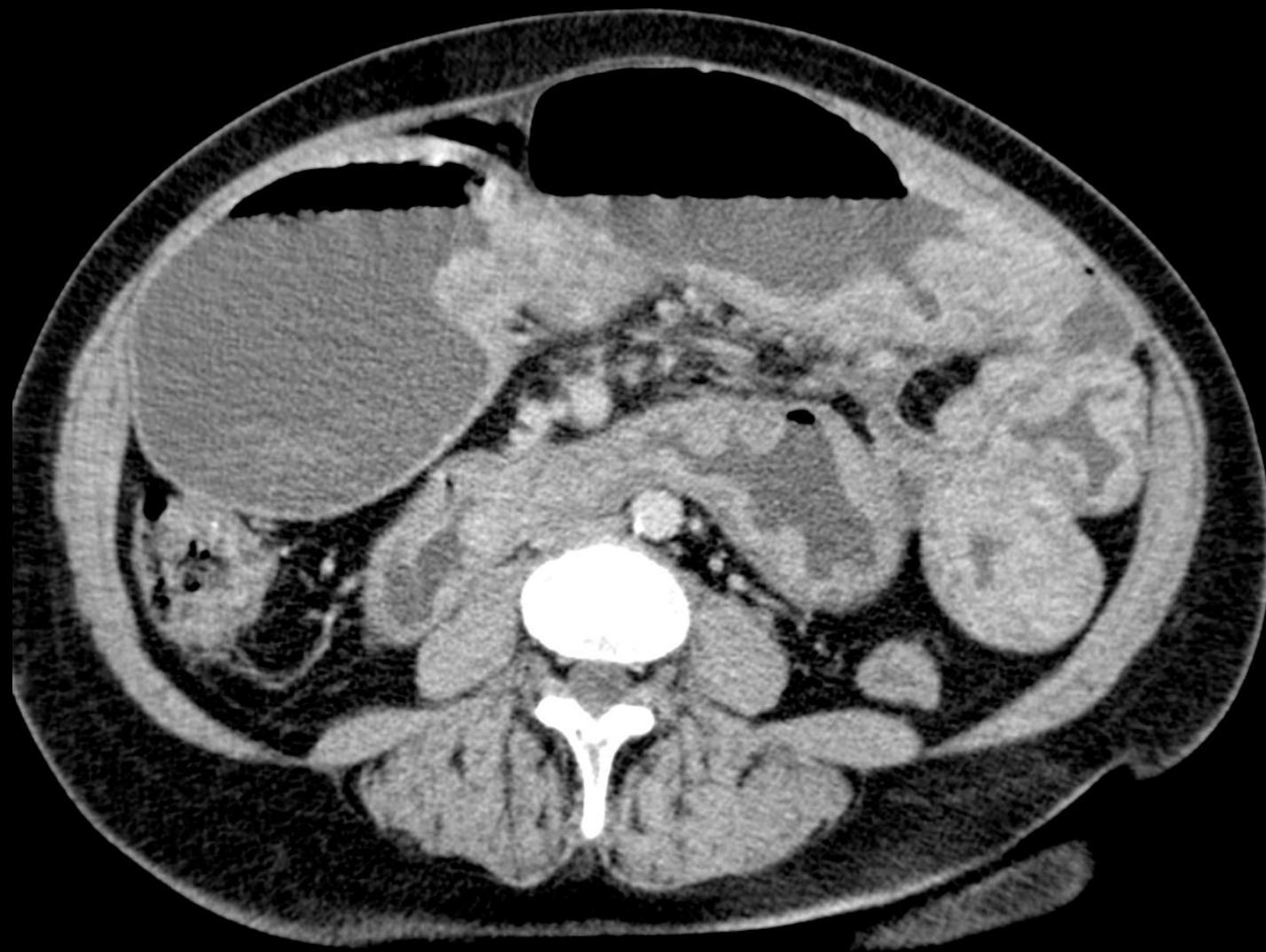


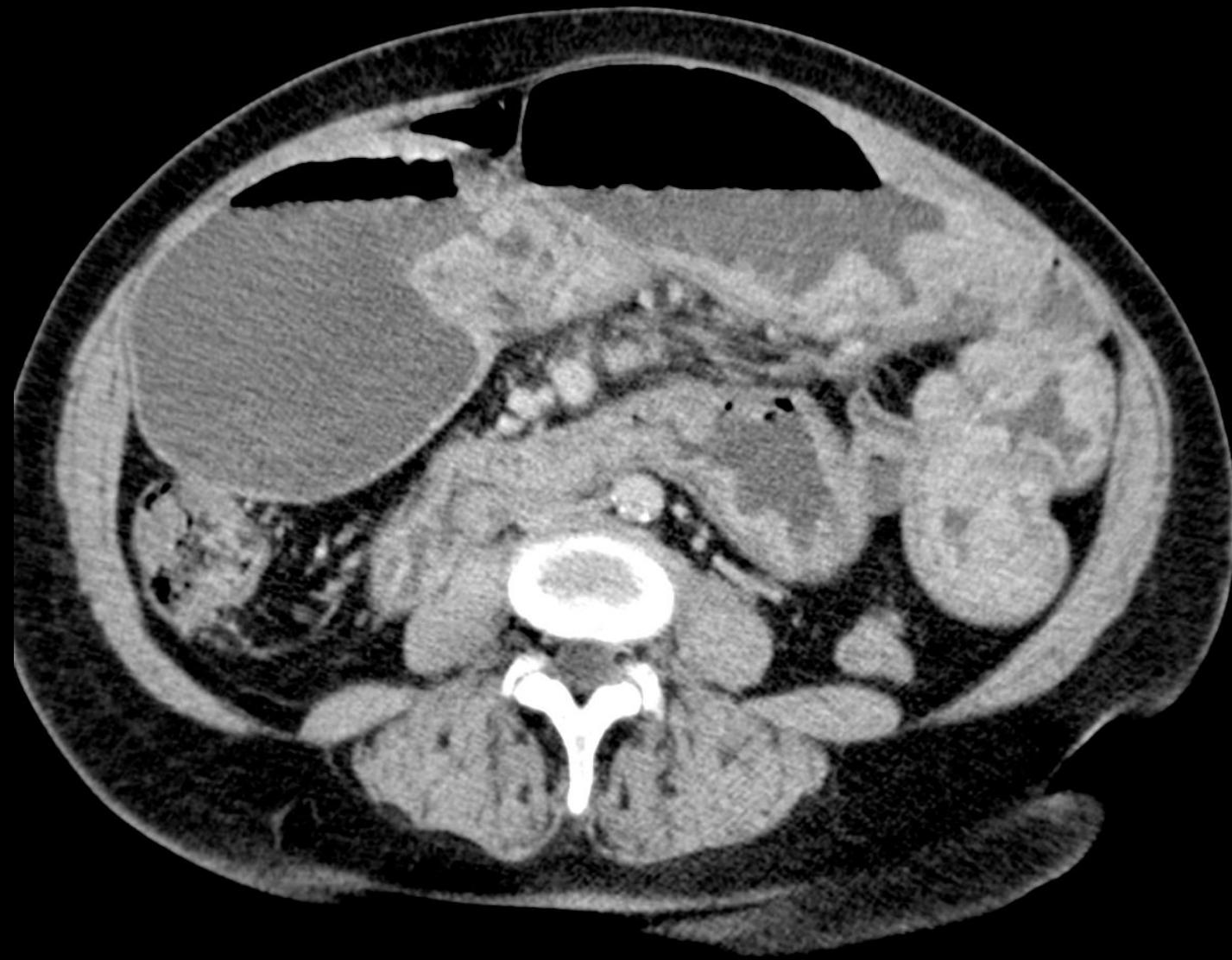


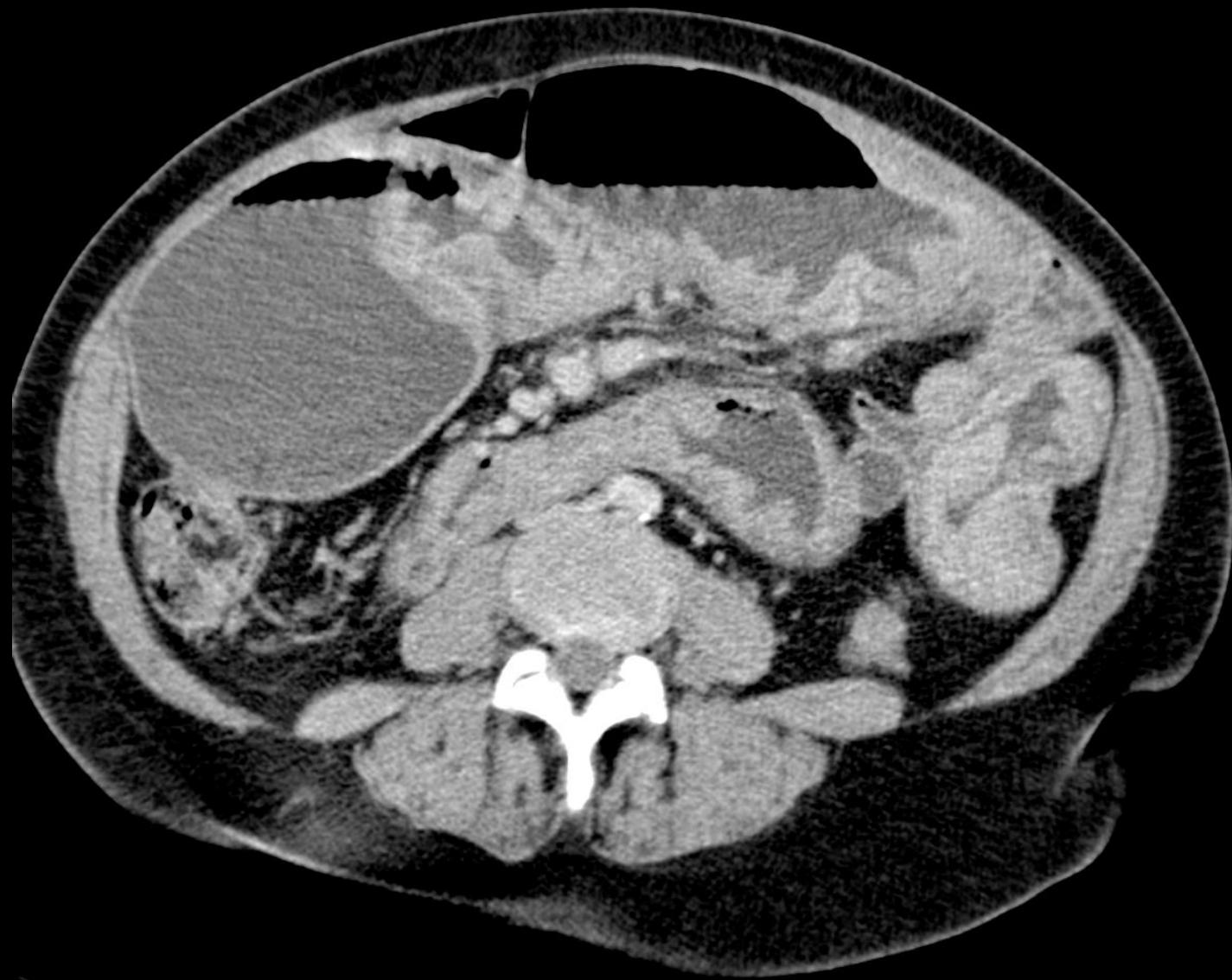


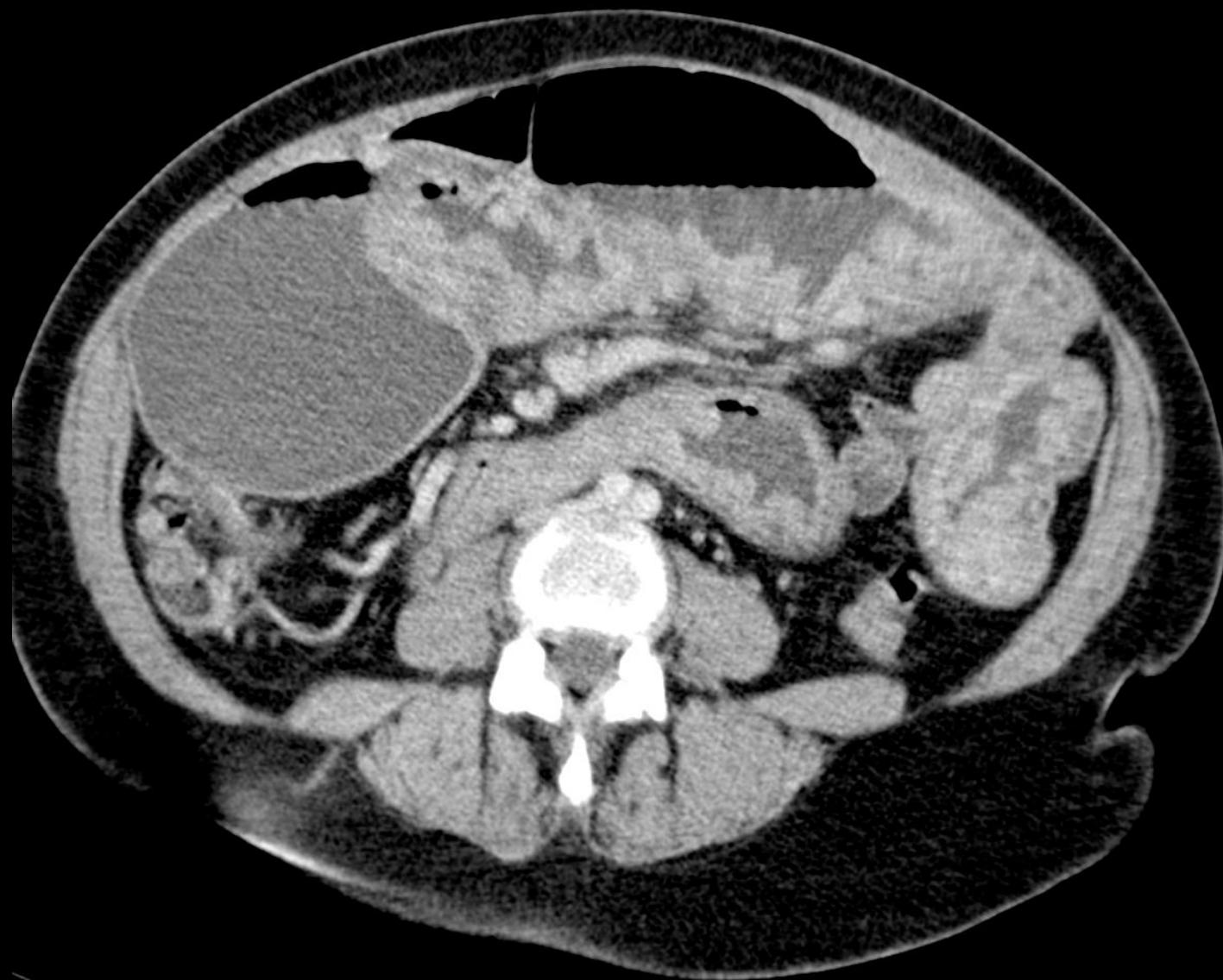


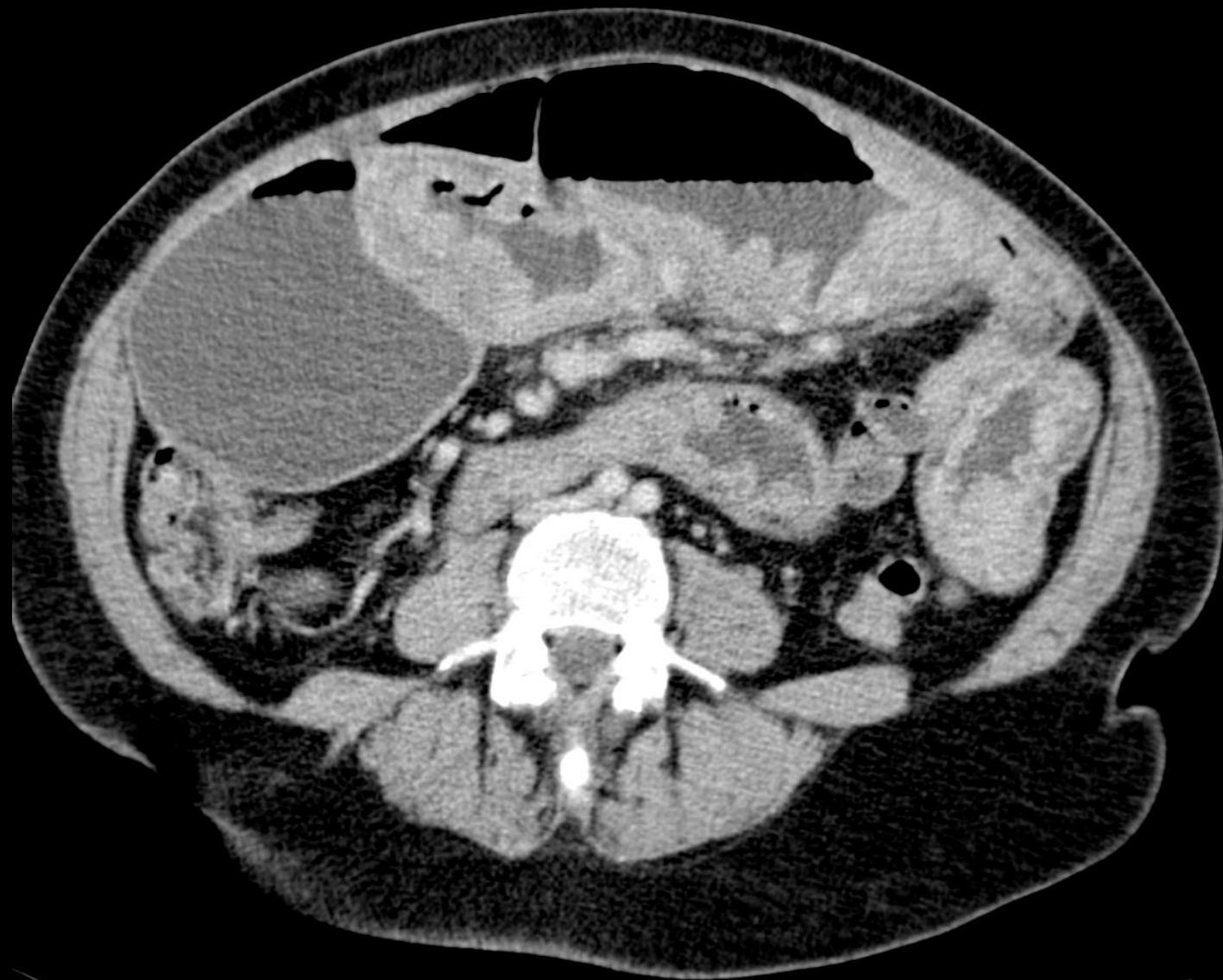


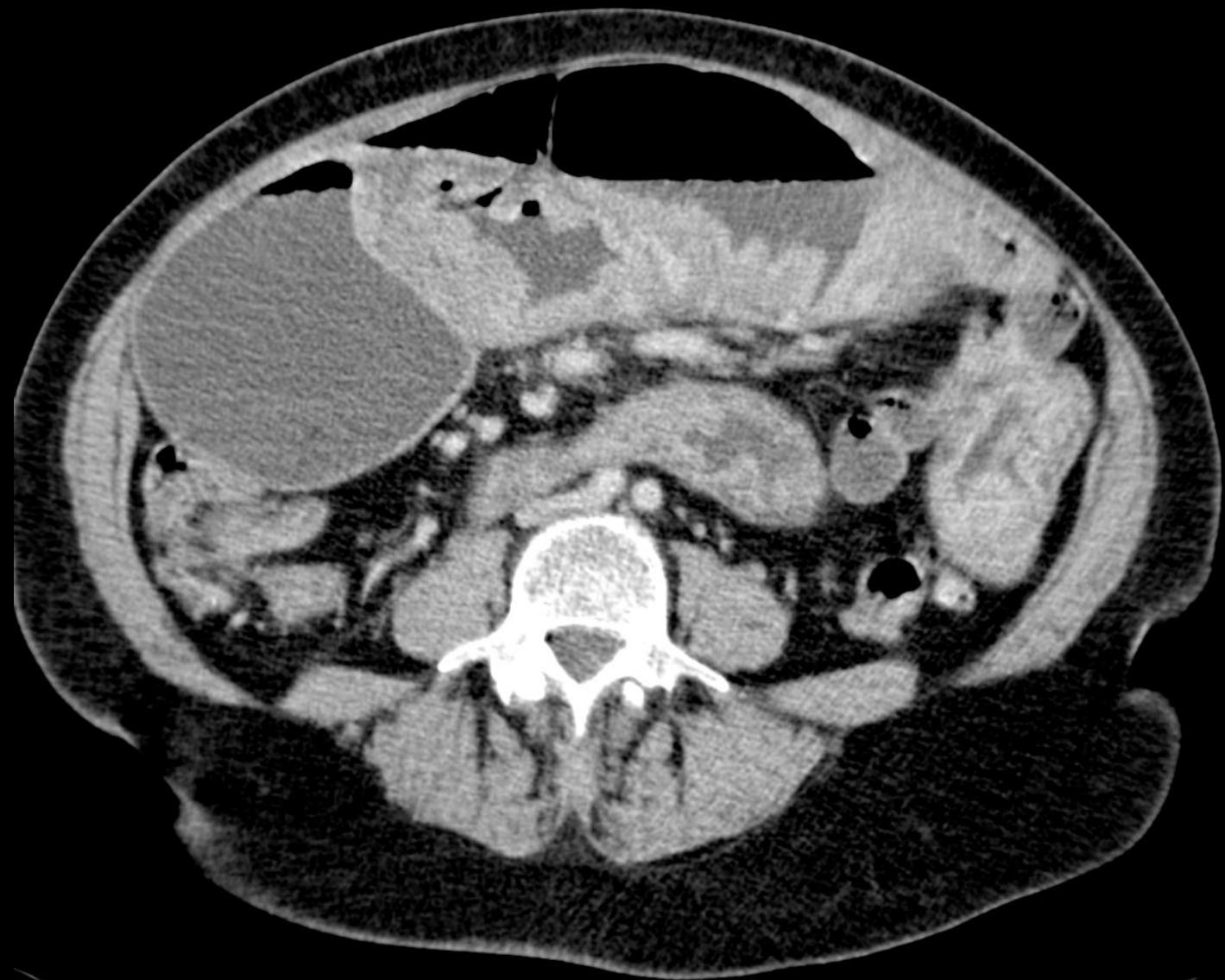


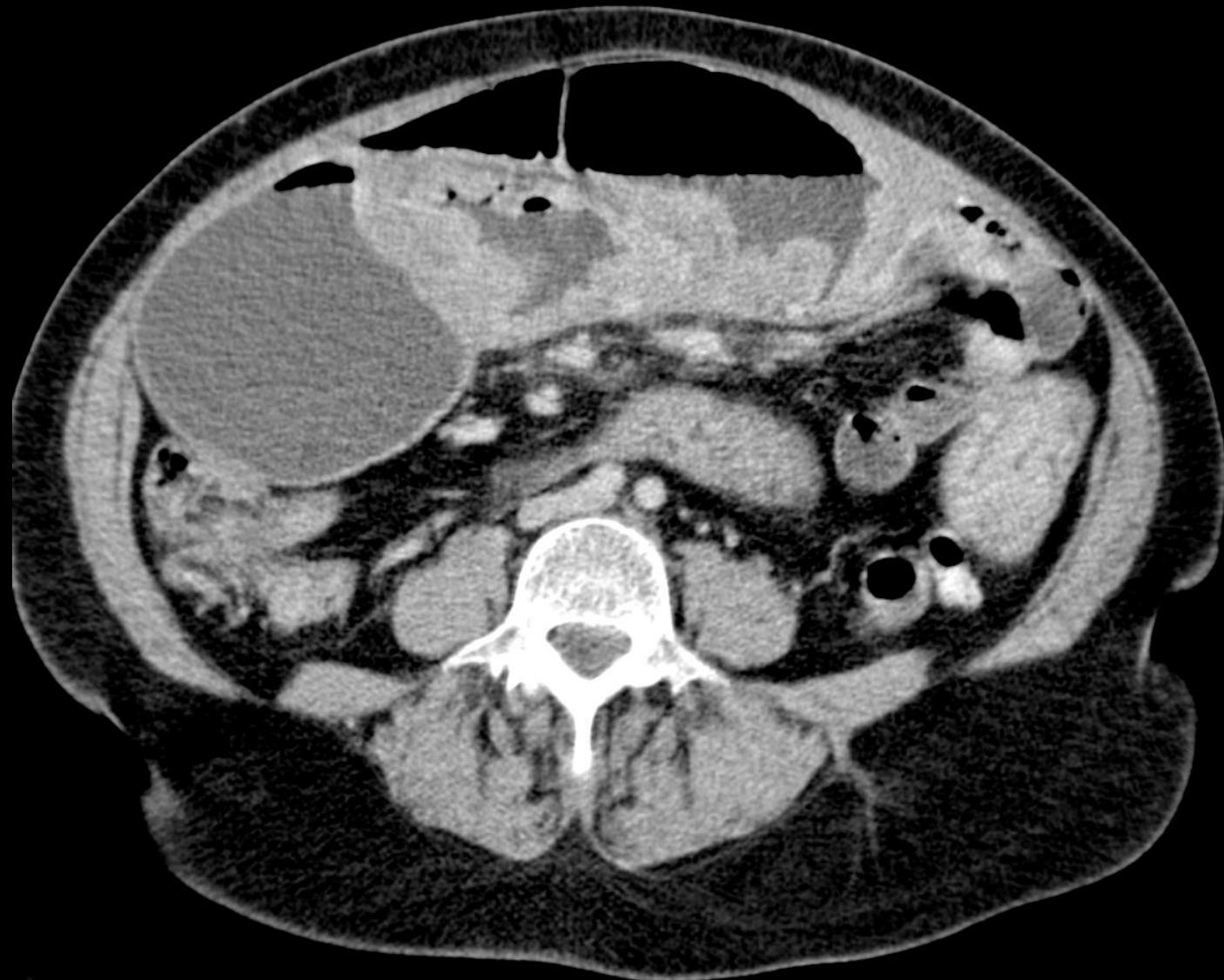


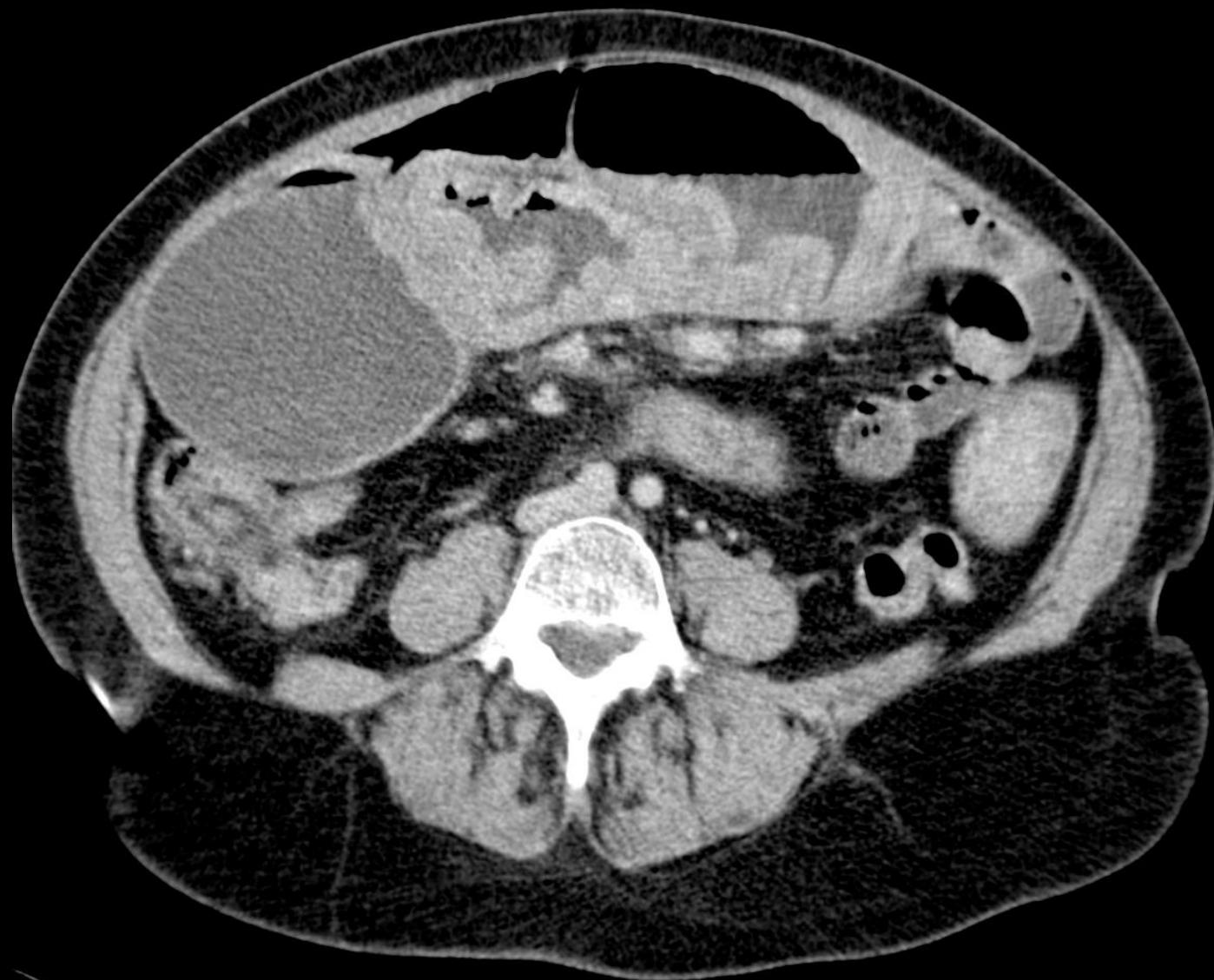


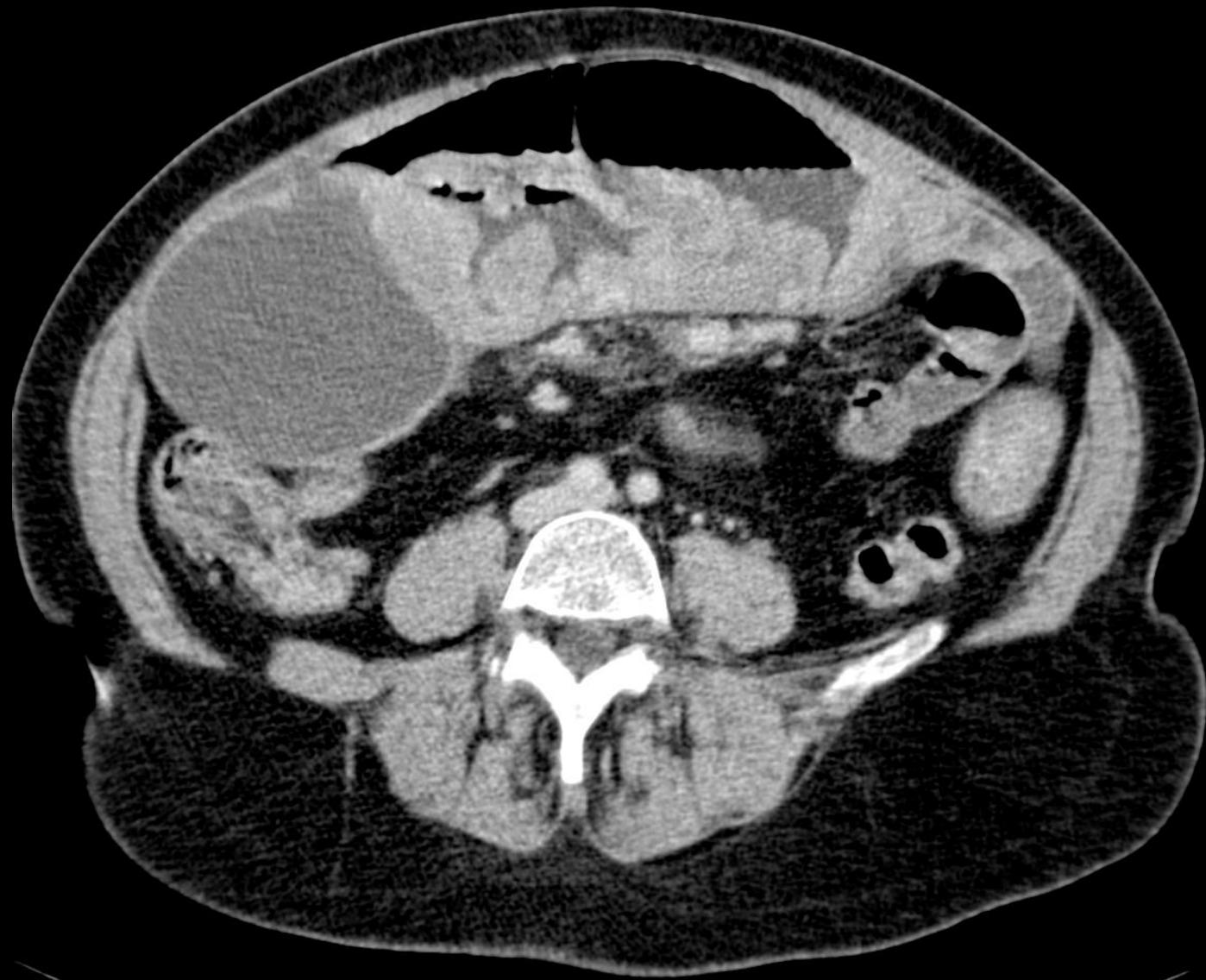


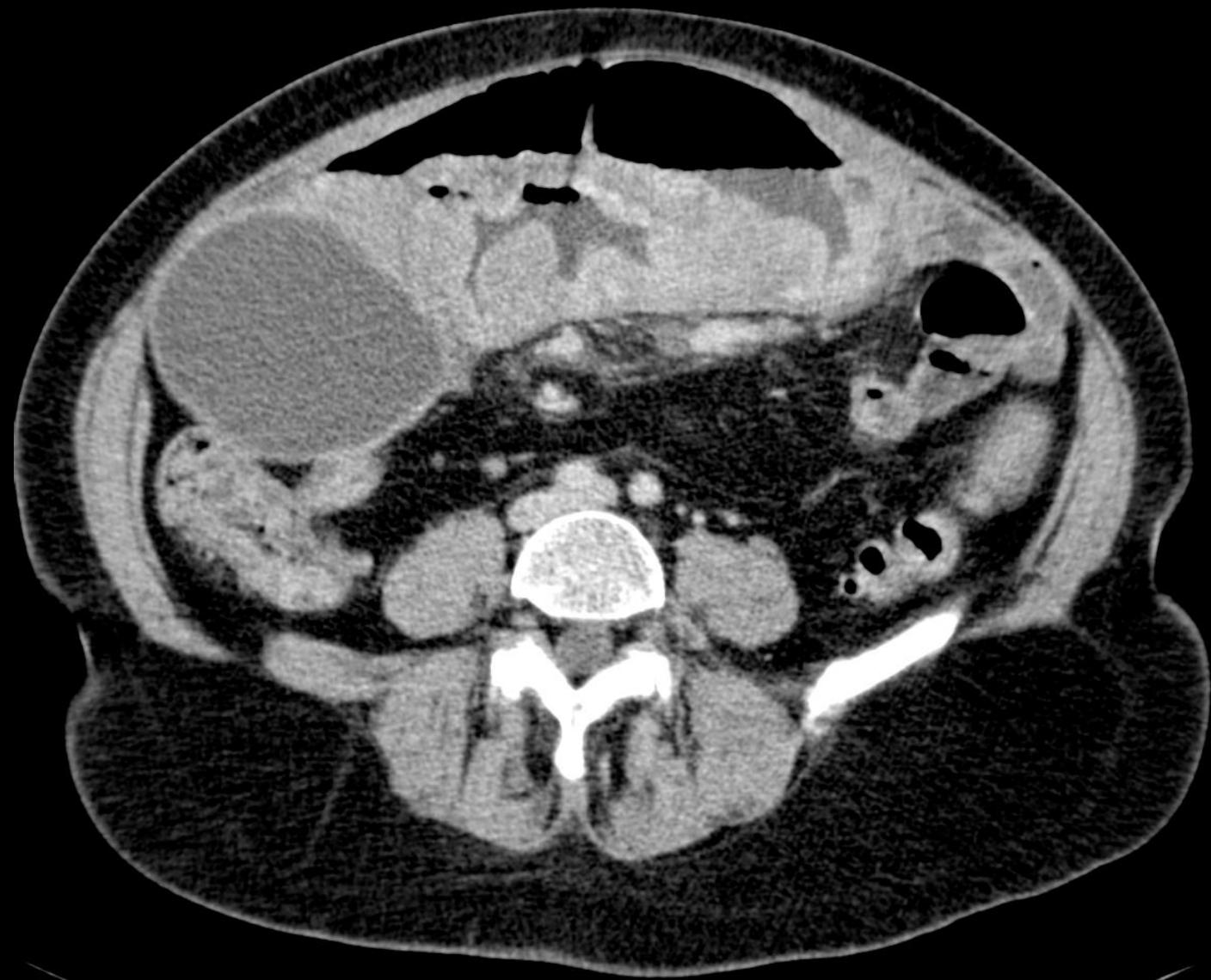


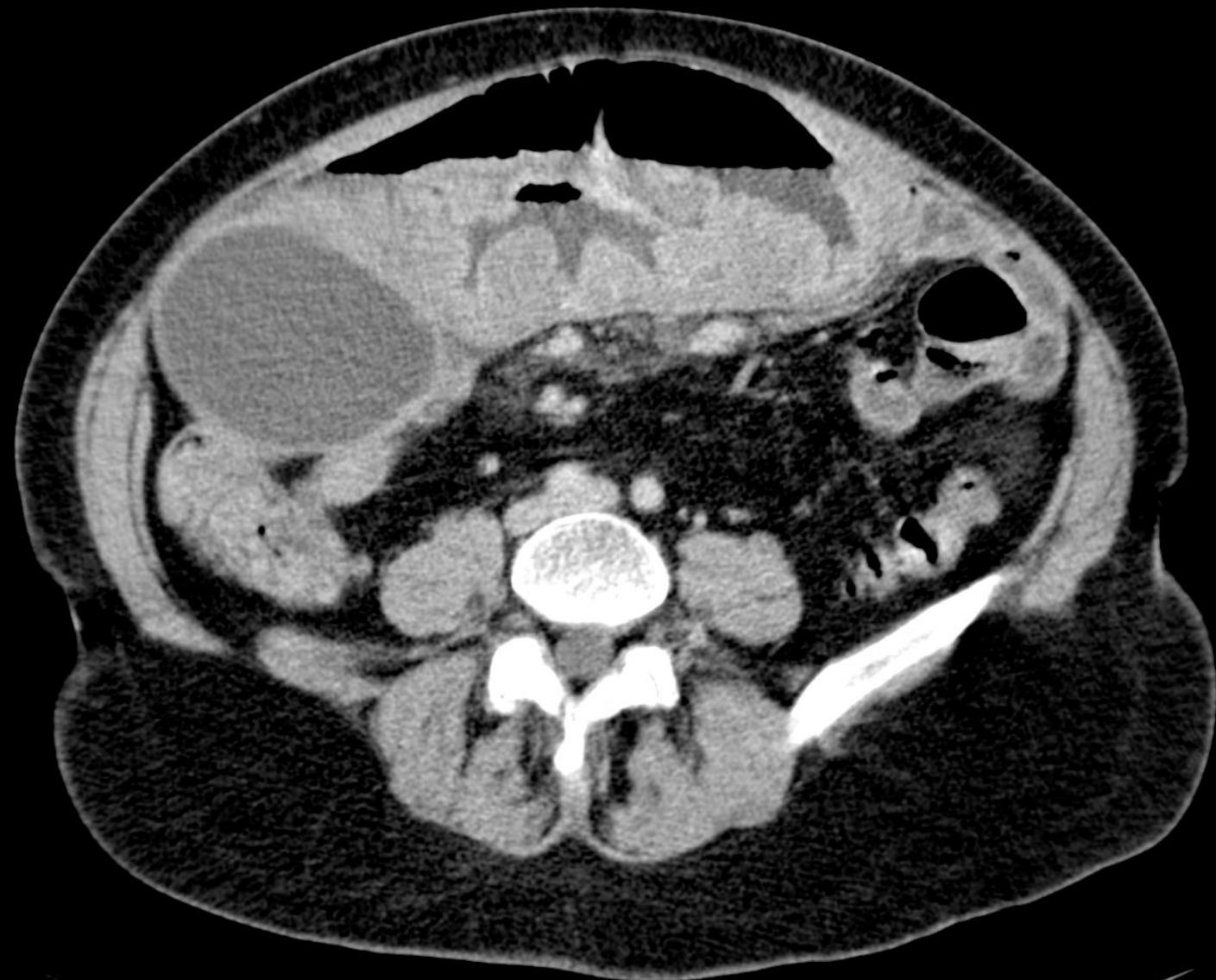


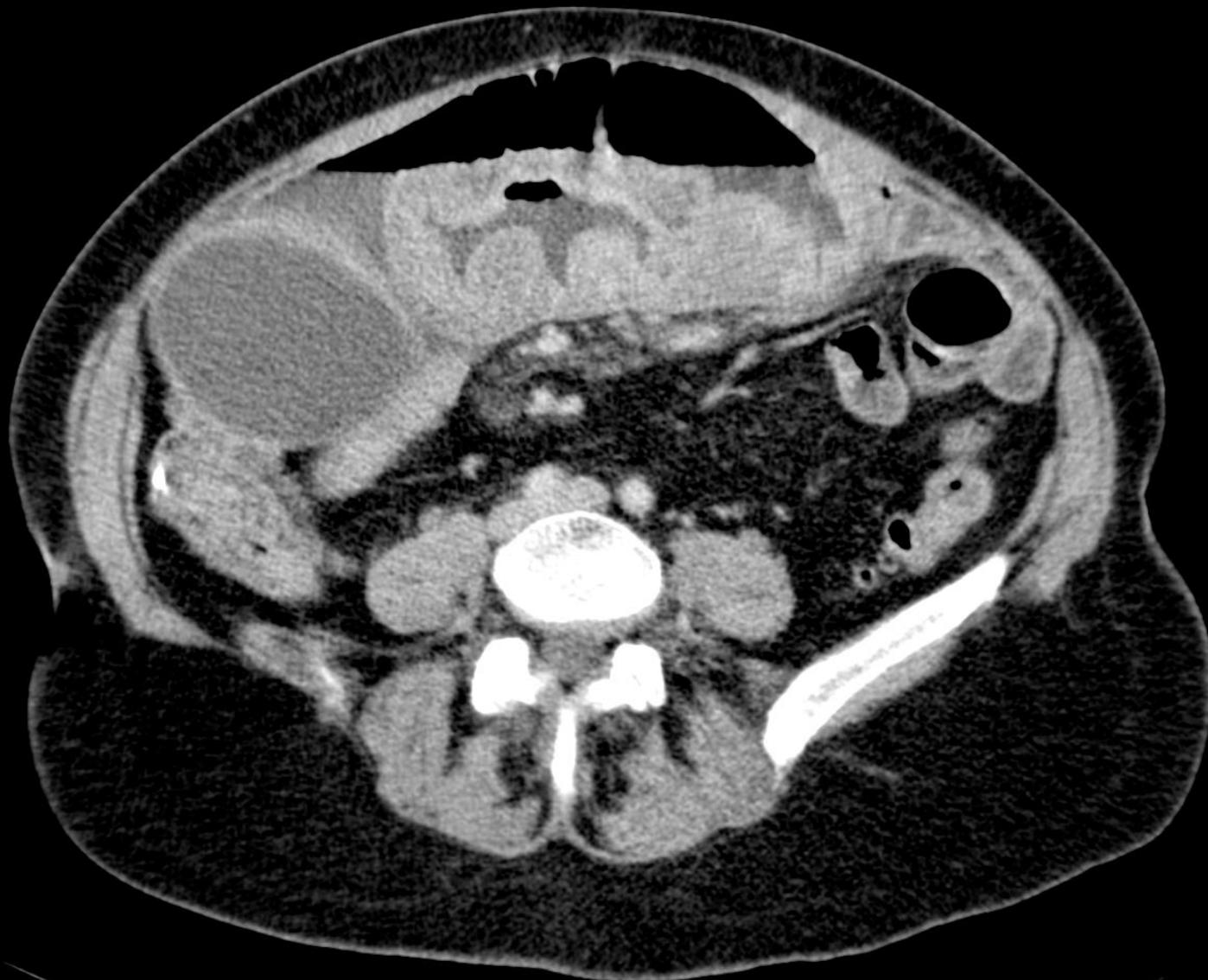


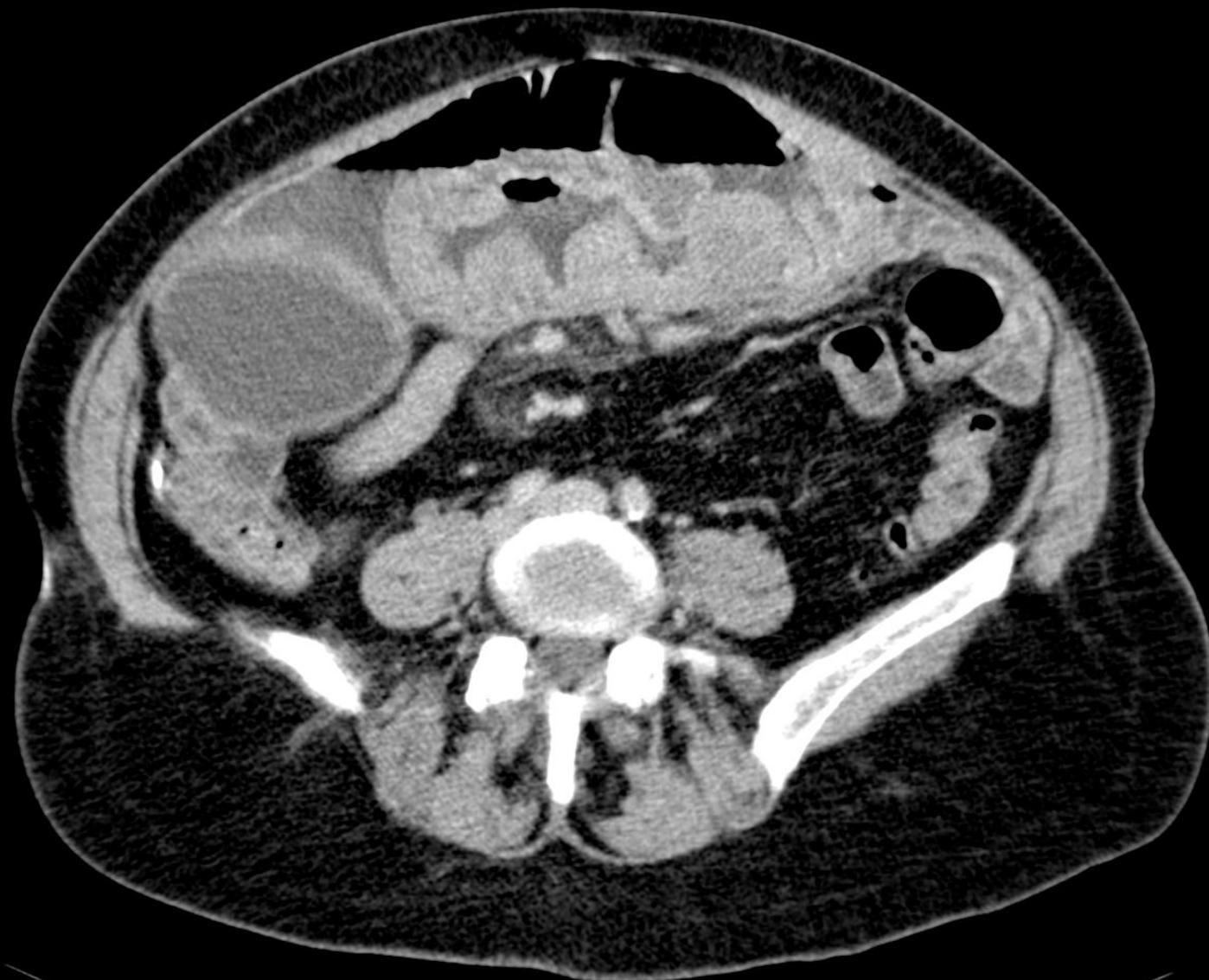


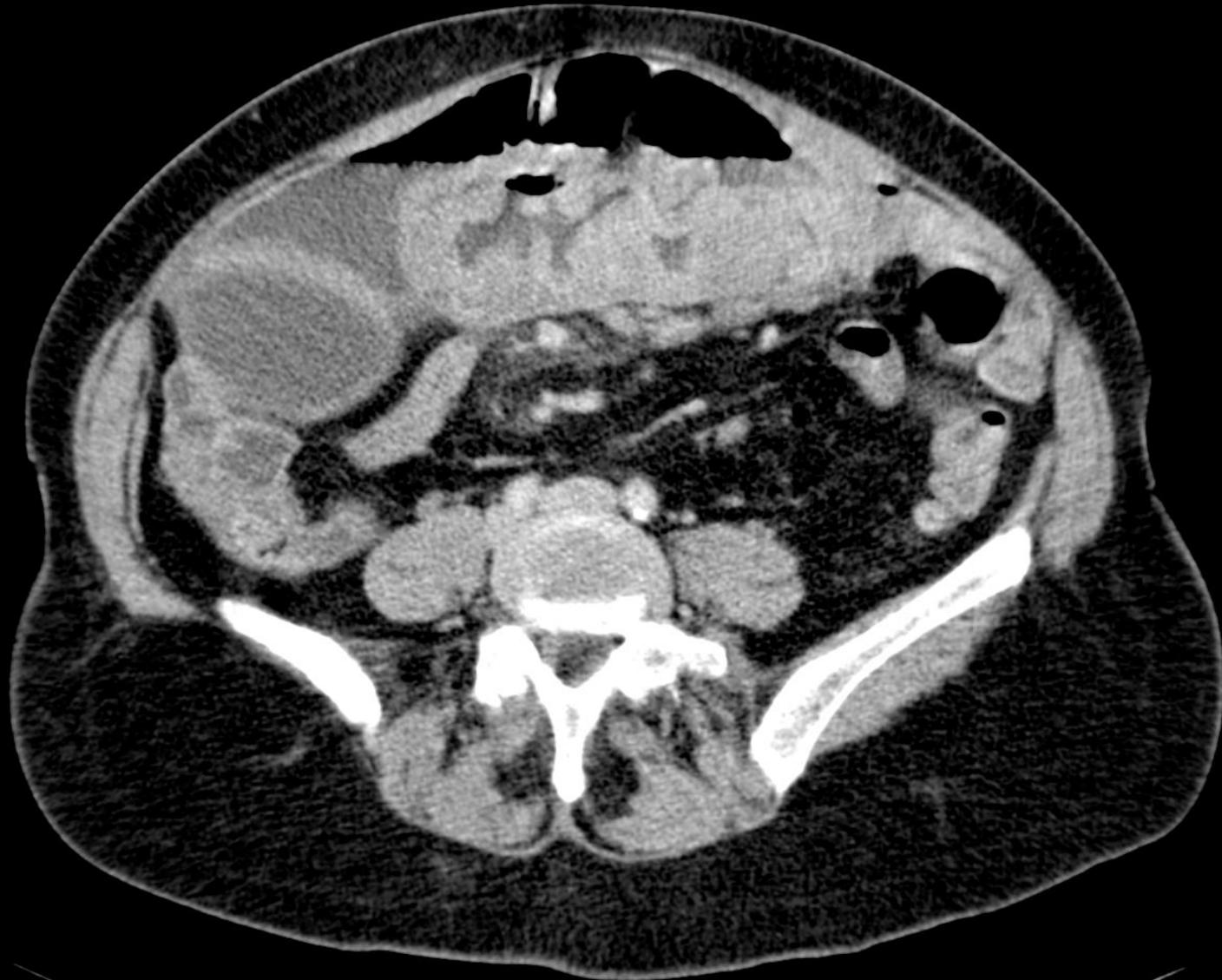






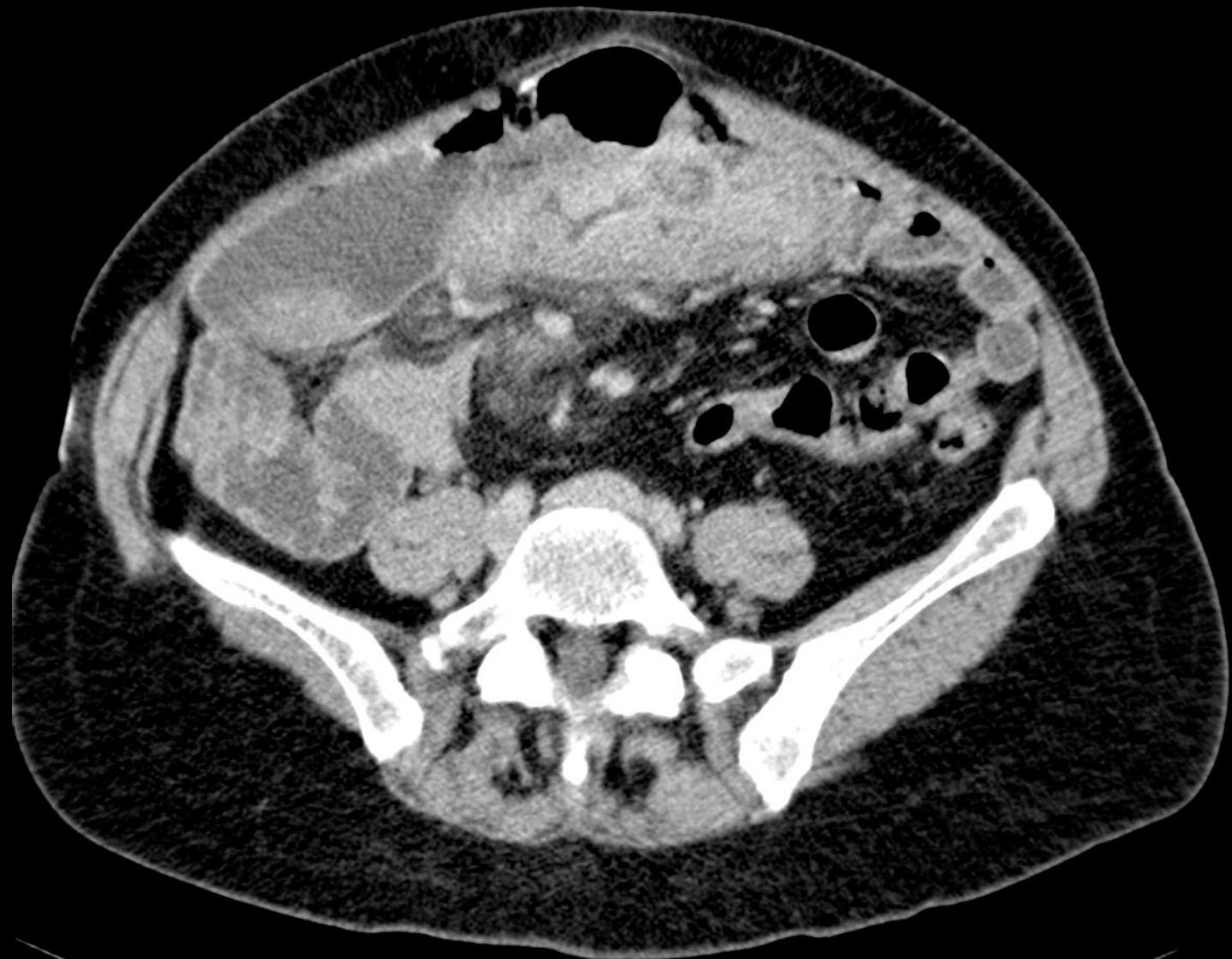




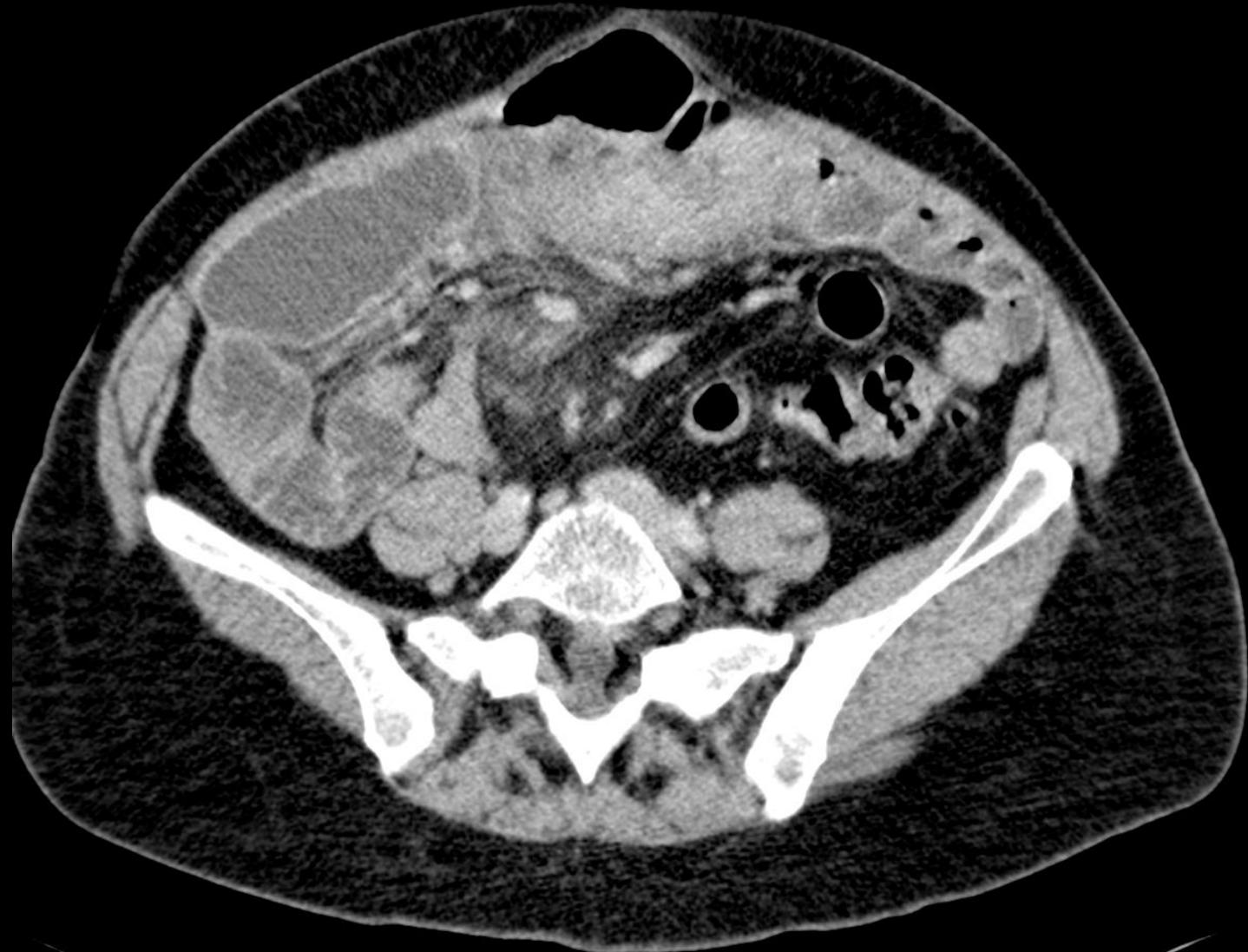










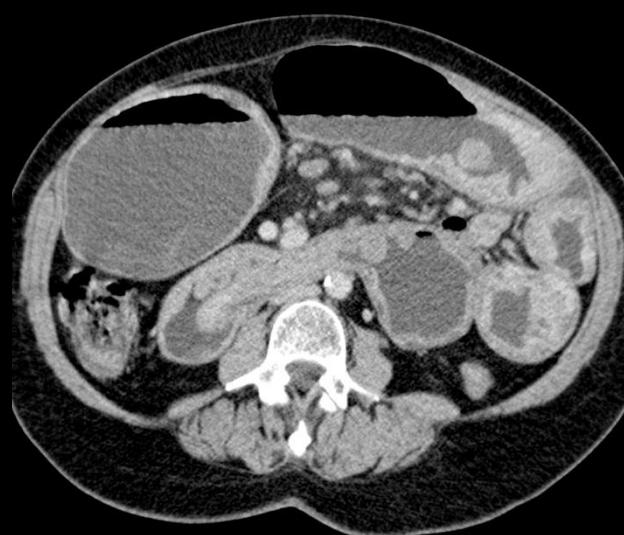
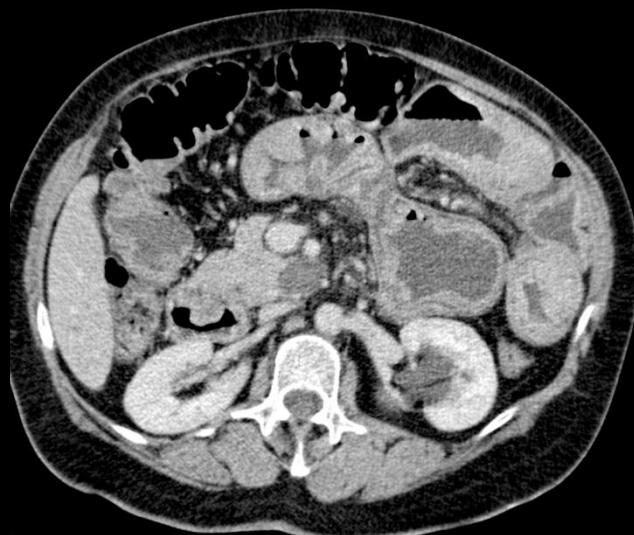
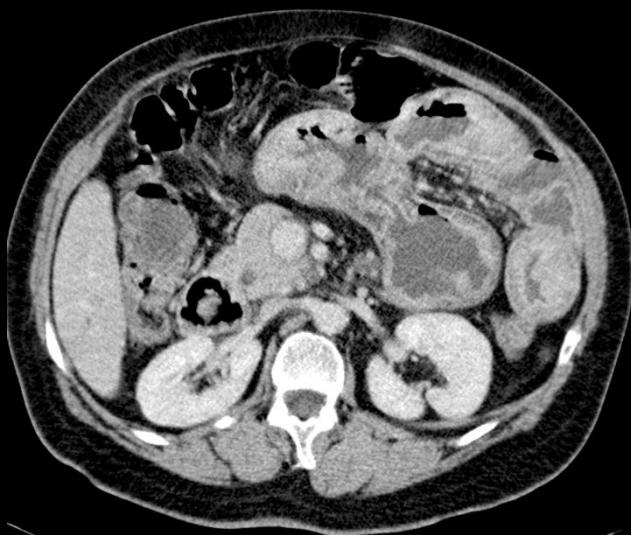




# Hipóteses Diagnósticas

♀ 54 anos

Distensão abdominal e enfartamento pós prandial





# Hipóteses Diagnósticas

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- Neoplasias
  - Linfoma, GIST, T. Neurogénico
- Síndromes Polipóides
  - Peutz-Jeghers
- Infecção
  - Yersinia, Giardia, Strongyloides, oportunística
- Enteropatia Eosinofílica, Amiloidose, Mastocitose, Macroglobulinemia Waldenstrom



# Espessamento pregas delgado

Espessamento das Pregas	Irregular	Regular
Segmentar	Linfoma DC Tuberculose Yersinia, Giardia, Strongyloides Infecção oportunistica	Isquemia Vasculite Radiação Hemorragia intramural
Difuso	Doença Whipple Linfangiectasia intestinal Gastroenterite Eosinofílica Mastocitose Amiloidose Macroglobulinemia Waldenstrom Infecções oportunisticas	Edema Enterite eosinofílica Abetalipoproteinemia Amiloidose



# Linfoma intestinal

- 1<sup>ário</sup> vs 2<sup>ário</sup> (+++)
- TGI: local extraganglionar + comum
- estômago › delgado (ileo distal – placas de Peyer)
- ↑ risco: d.celiaca, SIDA, LES, DC, QT

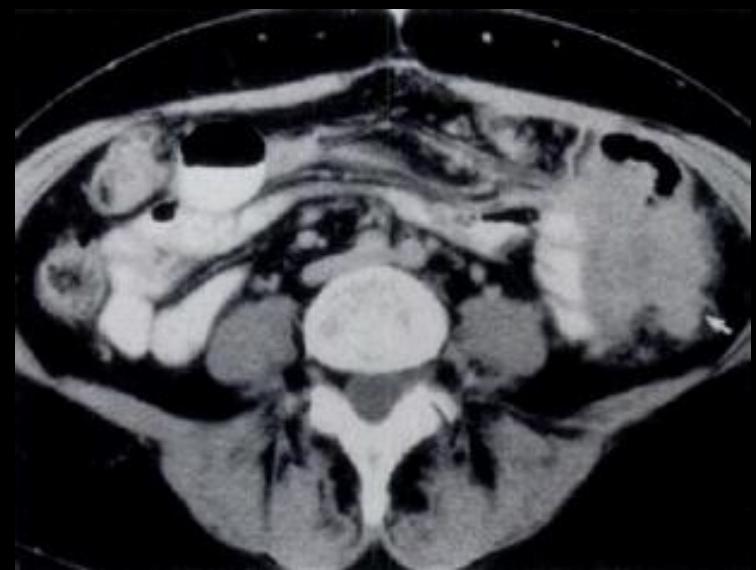
Espressamento nodular  
Pequenos pólipos  
Lesão infiltrante  
Massa excêntrica





# Linfoma intestinal

- Hepatoesplenomegalia, adenopatias
- *Sandwich sign*
- Dilatação aneurismática
- Ulceração e necrose
- Multifocal 10 – 25%





# GIST (Gastrointestinal Stromal Tumors)

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- 0,1-3% tumores TGI ; 5,7% sarcomas
  - 70-80% benignos
  - > 50 anos
- 
- Precursors das cél. intersticiais de Cajal
  - Expressão *KIT protein* (CD117)
- 
- TGI (estômago), mesentério
  - Muscular própria, muscular mucosa



# GIST

- Componente exofítico +++  
(obstrução não comum)
- Endoluminal (polipóide)

Necrose central, c/s/NHA  
Ulceração e hemorragia  
Obstrução não comum  
Raramente múltiplos (maligno)





# GIST

Benigno/Maligno:

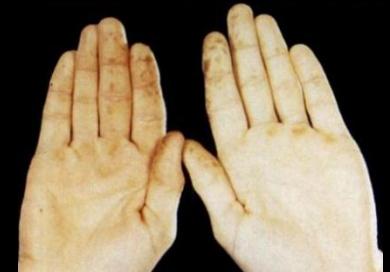
- > 5 cm
- Necrose e hemorragia
- Hipercelularidade
- Atipia nuclear
- ↑ índice mitótico

- > 5 cm
- Realce heterogéneo
- Metástases (fígado e peritoneu)
- Componente cístico-necrótico

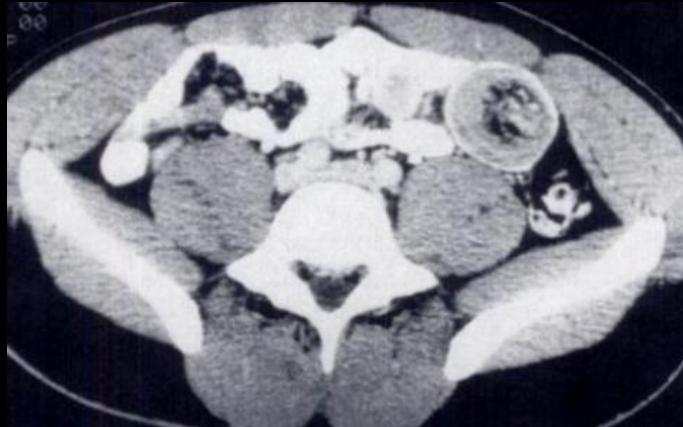


# Síndrome Peutz-Jeghers

- Polipose intestinal hereditária AD (19p13.3)
- 3ª década de vida



Syndrome	Distribution of Polyps	Histologic Type	Malignant Potential	Associated Lesions
Peutz-Jeghers syndrome	Small and large intestines, stomach	Hamartoma	Rare	Mucocutaneous pigmentation; tumors of the ovary, breast, pancreas, endometrium





# Outros

Gastroenterite eosinofílica  
(eosinófilos)

Geralmente patologia gástrica associada  
Eosinofilia periférica (75%) e atopia (50%)  
Mucosa(diarreia), transmural(obstr), serosa (ascite)

Amiloidose  
(proteína fibrilar insolúvel)

Sist GI (colon, delg 70%)  
Espess pregas (nod /dif), adp, hepatoesplenomegalia  
↓ motilidade (destruição plexo mioentérico); dilatação

Mastocitose  
(mastócitos)

Flush, prurido, cefaleias, sintomas GI (histamina)  
Nodularidade mucosa difusa

Macroglobulinemia Waldenstrom  
(imunoglobulina M)

Hepato-esplenomegalia, adenopatias  
Micronód.(1-2mm),adp,hepatoesplenomeg,ascite,dens.mesentério

D.Whipple (*Tropheryma whipplei*)  
(macrófagos PAS +)

Malabsorção, febre, artralgia  
Jejuno:padrão mucoso discretamente nodular, lúmen normal  
Adenopatias mesentéricas e retroperit.(hipodensas)  
Hepatoesplenomeg, ascite, ausência de dilatação

Linfangiectasia Intestinal

1ária (D.Milroy) / 2ária (inflam/neoplásica), generalizada/localizada  
Espess nod dif (linfáticos dilatados)  
Padrão mucoso granular grosso

Infecção

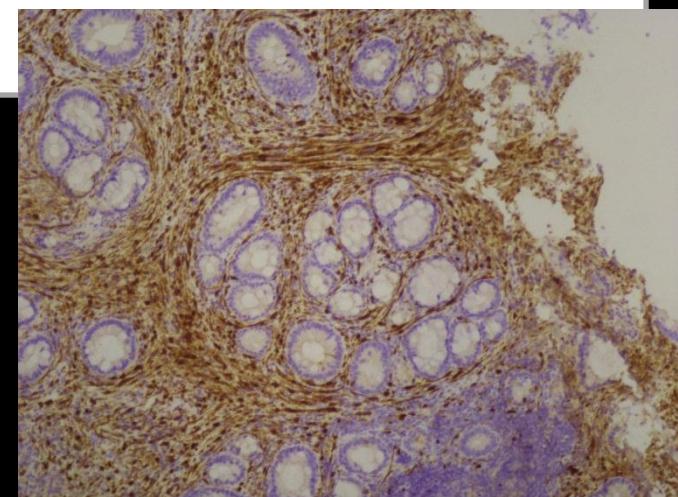
Giardia, Yersinia, Strongyloides, oportunística



# Anatomia Patológica

## Diagnóstico Histopatológico

1. Duodeno distal com proliferação na lâmina própria de fibras nervosas e células ganglionares, enquadrável em **polipose ganglioneuromatosa** (correlação clínico-patológica).
2. Aspectos reactivos do tipo das vilosidades do duodeno distal.



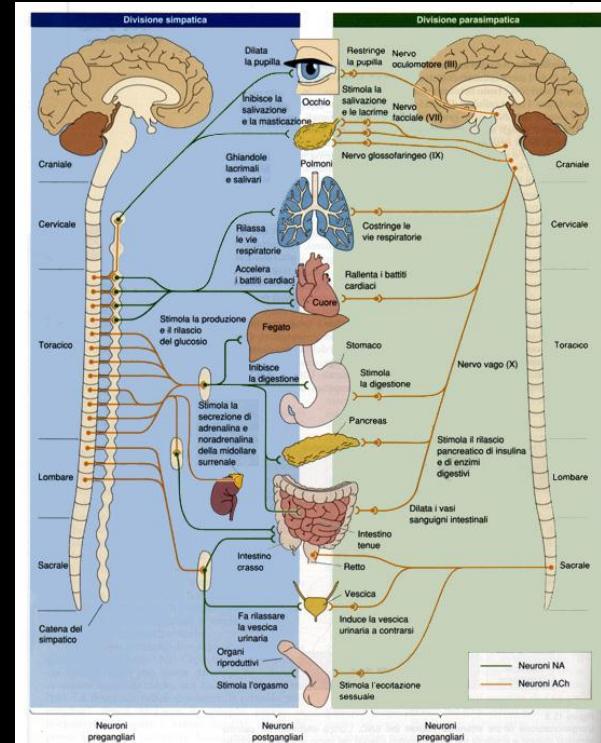
S100 e NSE +

# T. Neurogénicos abdominais

- Células Ganglionares
  - Ganglioneuromas
  - Ganglioneuroblastomas
  - Neuroblastomas

- Sistema Paraganglionar
  - Feocromocitoma
  - Paraganglioma

- Baínha Nervosa
  - Neurofibromas
  - Neurilemomas
  - Neurofibromatose
  - T. malignos baínha céls nervosas

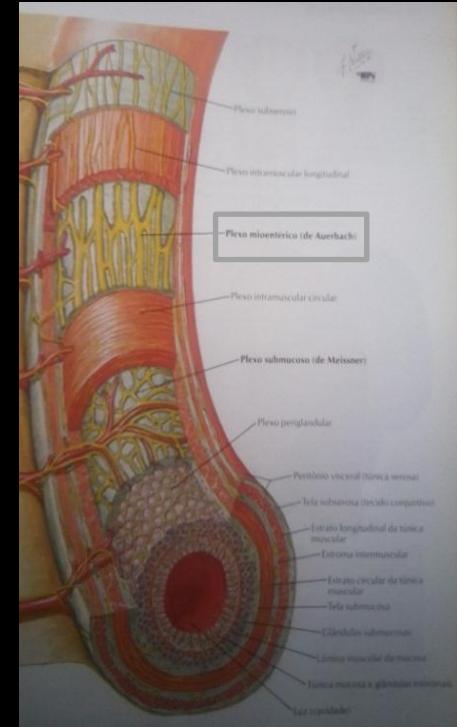


- Cadeia simpática paravertebral
- SR
- Órgão Zuckerkandel
- Outros (parede intestinal ,abd,vesícula, bexiga)



# Ganglioneuromatose intestinal

- Rara
- Hiperplasia plexo mioentérico
- Alterações da motilidade (obstipação)
- Produção de substâncias  
(VIP, catecolaminas, androgeneos)





# Ganglioneuromatose intestinal

Lesão polipóide focal

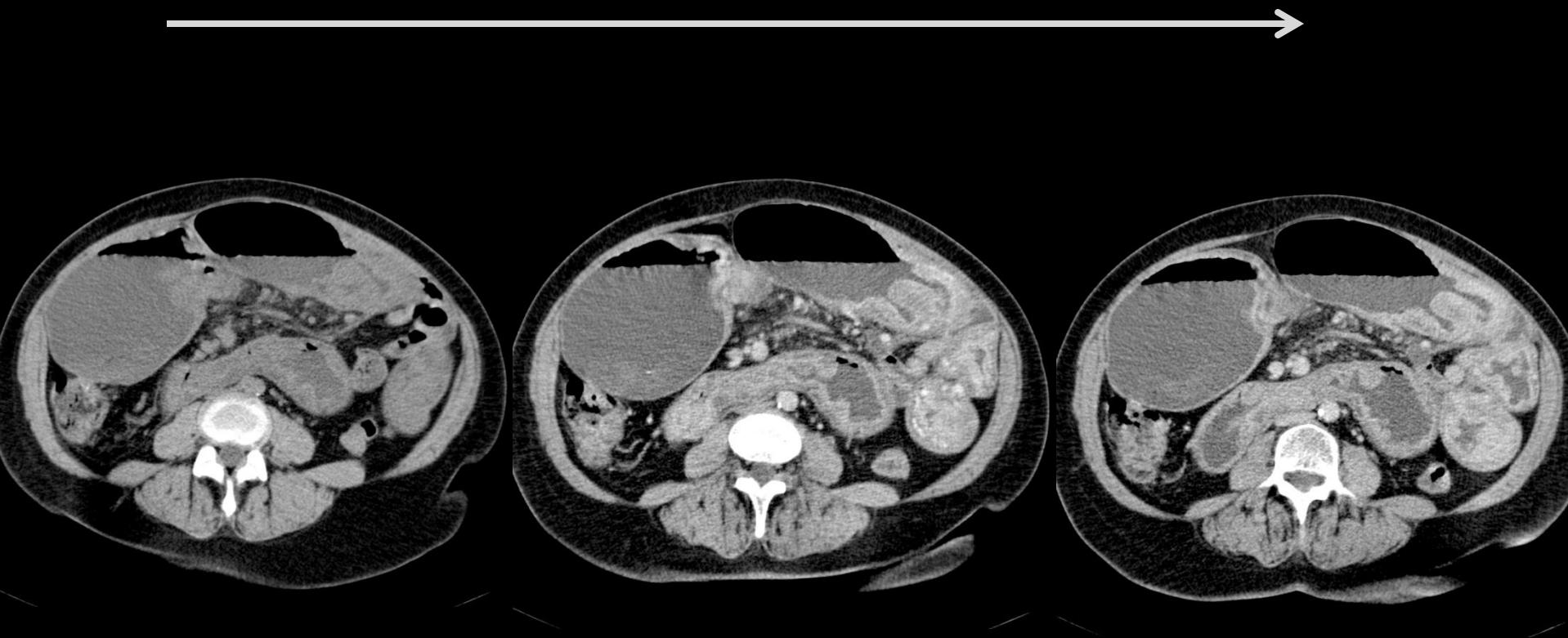
Polipose ganglioneuromatosa

Ganglioneuromatose difusa

→      colón  
          íleo terminal  
          apêndice

- Acumulação progressiva de contraste (matriz mixóide)

RadioGraphics 2003;23:29–43



Pré-contraste

Fase pancreática

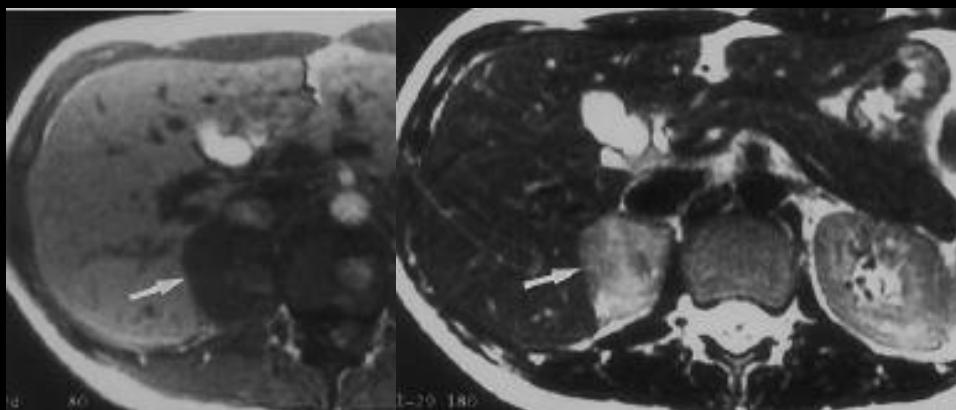
Fase portal



# Ganglioneuromas

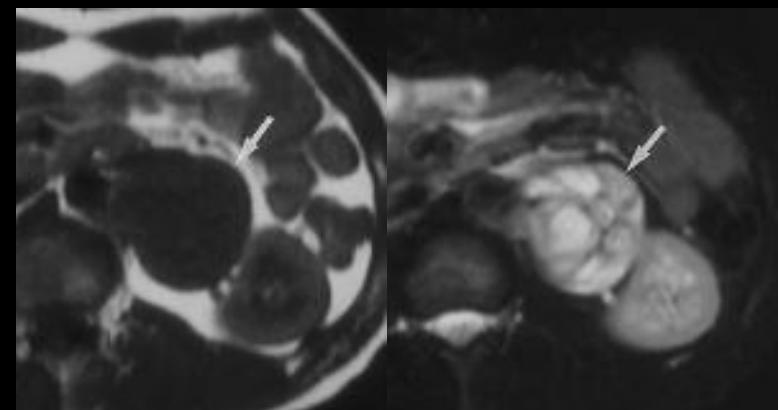
## RM

- T1: Homogéneo; intensidade de sinal baixa
- T2: Sinal variável
  - proporção estroma mixóide, componente celular, fibras de colagénio
  - bandas curvilíneas de hiposinal (cels Schwann e fibras de colagénio)



Ganglioneuroma SR (♀24A)

T1: homogénea, hipointenso .    T2: heterogénea, hiperintensa.



Ganglioneuroma retroperit. (♂20A)

T1: homogénea, hipointenso.T2: heterogénea (áreas lineares e curvilineas de hiposinal), marcadamente hiperintensa.



# Ganglioneuromatose intestinal

- Geralmente associada a MEN2b ou NF1
- Outras: ET, D.Cowden, Polipose juvenil, c.côlon
- Forma isolada extremamente rara



Síndrome MEN 2

Gene RET  
(Cr 10q; AD)

Subtipo 2A; Hiperparatiroidismo primário; FEO; carcinoma medular da tireoide.  
Subtipo 2B: Carcinoma medular da tireoide; FEO; ganglioneuromas; *habitus marfanóide*.

Neurofibromatose tipo 1

Gene NF-1  
(Cr 17q; AD)

Manchas cutâneas café-au-lait; neurofibromas; FEO; gliomas do nervo óptico; hamartomas da íris.





# Exames Auxiliares de Diagnóstico

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Despiste de outras patologias associadas



Negativo

# Ganglioneuromatose intestinal isolada

## Diffuse ganglioneuromatosis with plexiform neurofibromas limited to the gastrointestinal tract involving a large segment of small intestine

KEIJI HIRATA,<sup>1</sup> KOHTARO KITAHARA,<sup>1</sup> YASUHIRO MOMOSAKA,<sup>2</sup> HIROMI KOUHO,<sup>3</sup> NAOKI NAGATA,<sup>1</sup> HIROSHI HASHIMOTO,<sup>3</sup> and HIDEAKI ITOH<sup>1</sup>

<sup>1</sup>First Department of Surgery and Departments of <sup>2</sup>Internal Medicine and <sup>3</sup>Pathology, University of Occupational and Environmental Health, 1-1 Iseigaoka, Yahatanishi-ku, Kitakyushu 807, Japan

### Isolated intestinal neurofibromatosis of colon. Single case report and review of the literature

M. Bononi  
A. De Cesare  
M.C. Stella  
E. Fiori  
G. Galati  
P. Agnelli  
M. Angelini  
A. Cimitan  
A. Lemos<sup>1</sup>  
V. Cangemi

*Isolated intestinal neurofibromatosis of the colon is a most unusual disease; from 1937 to 1989 only 12 cases have been reported. The differential diagnosis and treatment of this lesion are very difficult. A review of the literature is made and personal experience in the diagnosis and treatment of a case in a 68-year-old female is described.*

*Digest Liver Dis* 2006;32:734-42

**Key words:** diagnosis; isolated intestinal neurofibromatosis; therapy

*J Clin Pathol* 2007;60:222 doi:10.1136/jcp.2006.036905

### Letter to the editor

### Adult transmural intestinal ganglioneuromatosis is not always associated with multiple endocrine neoplasia or neurofibromatosis: a case report

Serena F Ledwidge<sup>1</sup>, Morgan Moorghen<sup>2</sup>, Rob J Longman<sup>3</sup>, Michael G Thomas<sup>3</sup>

### CLINICAL CASE SEMINARS

### Germline RET Codon 918 Mutation in Apparently Isolated Intestinal Ganglioneuromatosis

CHARIS ENG\*, DEBBIE J. MARSH, BRUCE G. ROBINSON, C. W. CHOW, MICHAEL A. PATTON, MELISSA C. SOUTHEY, DEON J. VENTER, BRUCE A. J. PONDER†, PETER J. MILLA, AND VIRPI V. SMITH

Online Submissions: wjg.wjgnet.com  
wjg@wjgnet.com  
doi:10.3748/wjg.14.6569



*World J Gastroenterol* 2008 November 14; 14(42): 6569-6571  
*World Journal of Gastroenterology* ISSN 1007-9327  
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### CASE REPORT

### Isolated intestinal neurofibromatous proliferations in the absence of associated systemic syndromes

J Elliot Carter, Javier A Laurini

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www.wjgnet.com  
wjg@wjgnet.com



*World J Gastroenterol* 2006 December 28; 12(48): 7874-7877  
*World Journal of Gastroenterology* ISSN 1007-9327  
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### CASE REPORT

### Sporadic ganglioneuromatosis of esophagogastric junction in a patient with gastro-esophageal reflux disorder and intestinal metaplasia

Richard Siderits, Iman Hanna, Zahid Baig, Janusz J Godyn



# Ganglioneuromatose intestinal isolada

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Tratamento → Caracterização pré-cx  
(localização, tamanho)



# Informação Clínica

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Agravamento das queixas

Vómitos

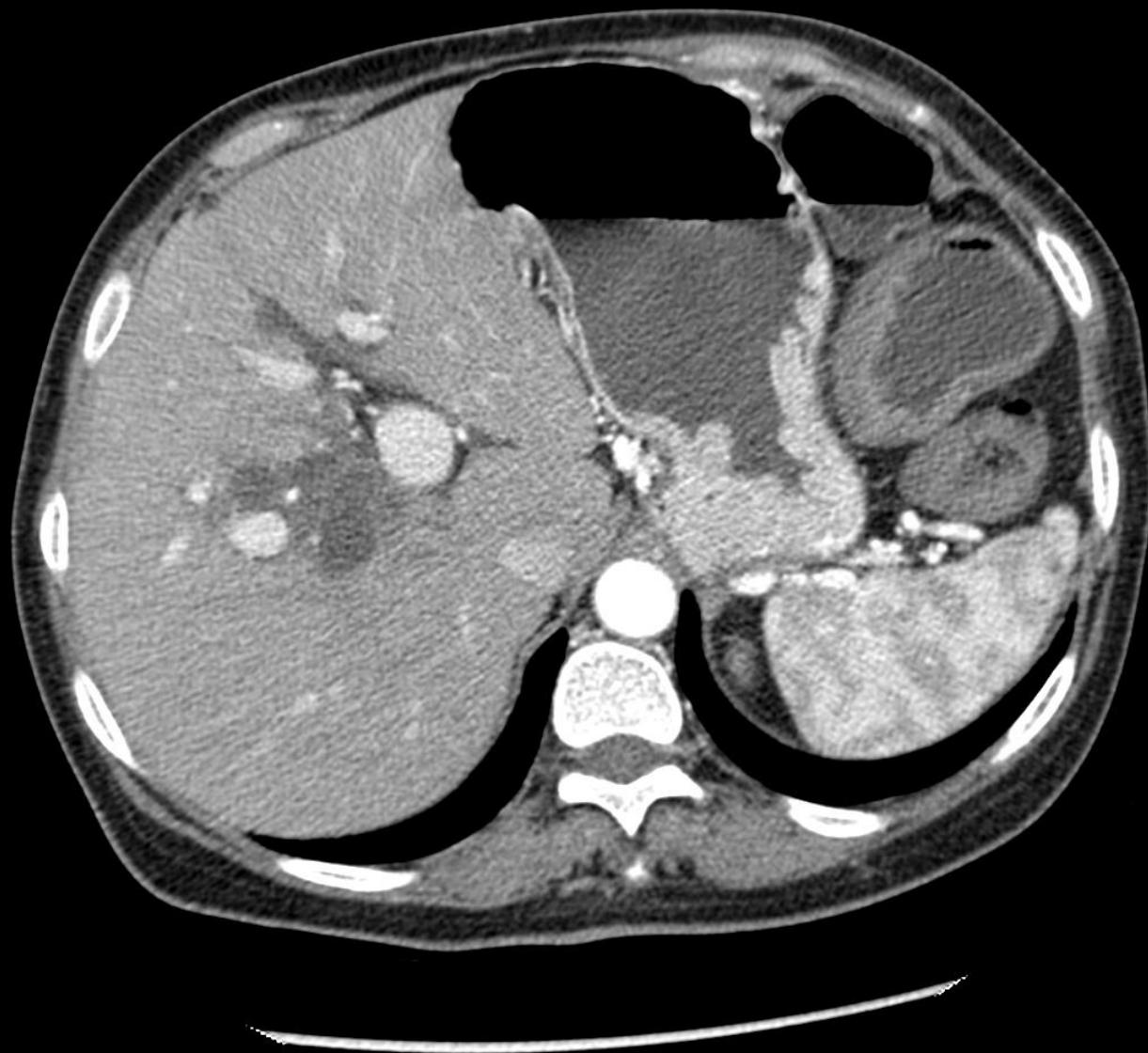
Diarreia

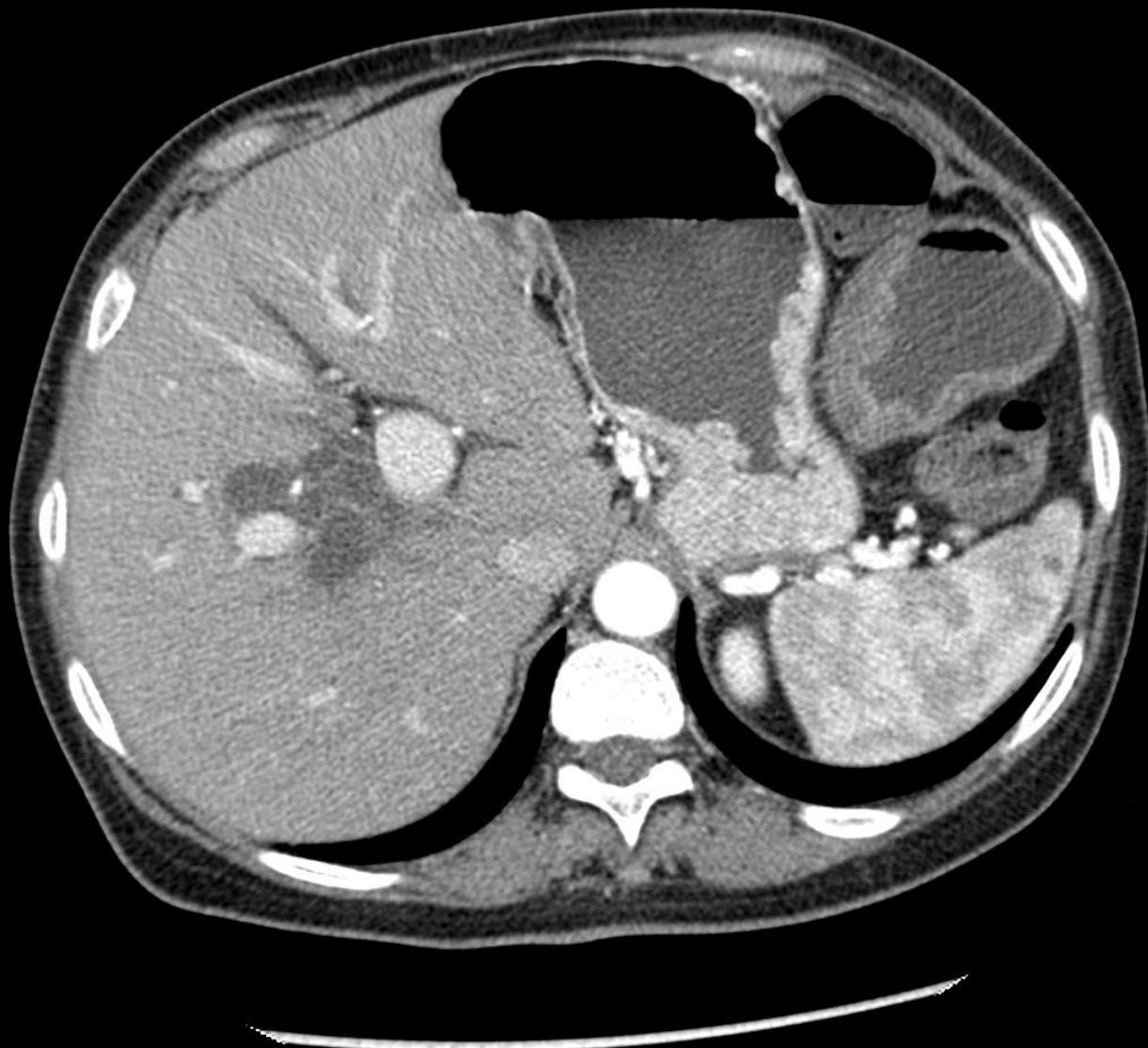
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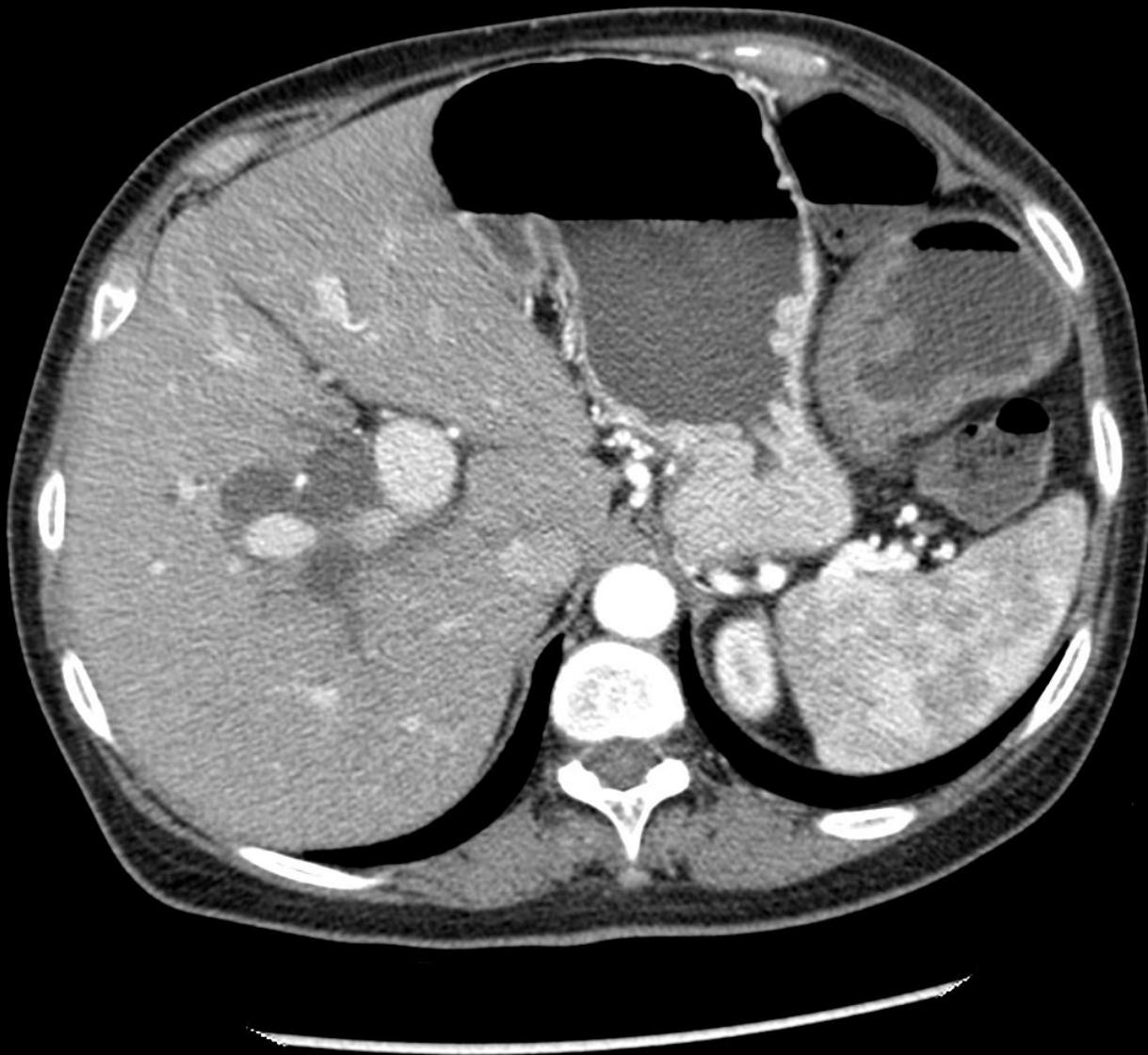


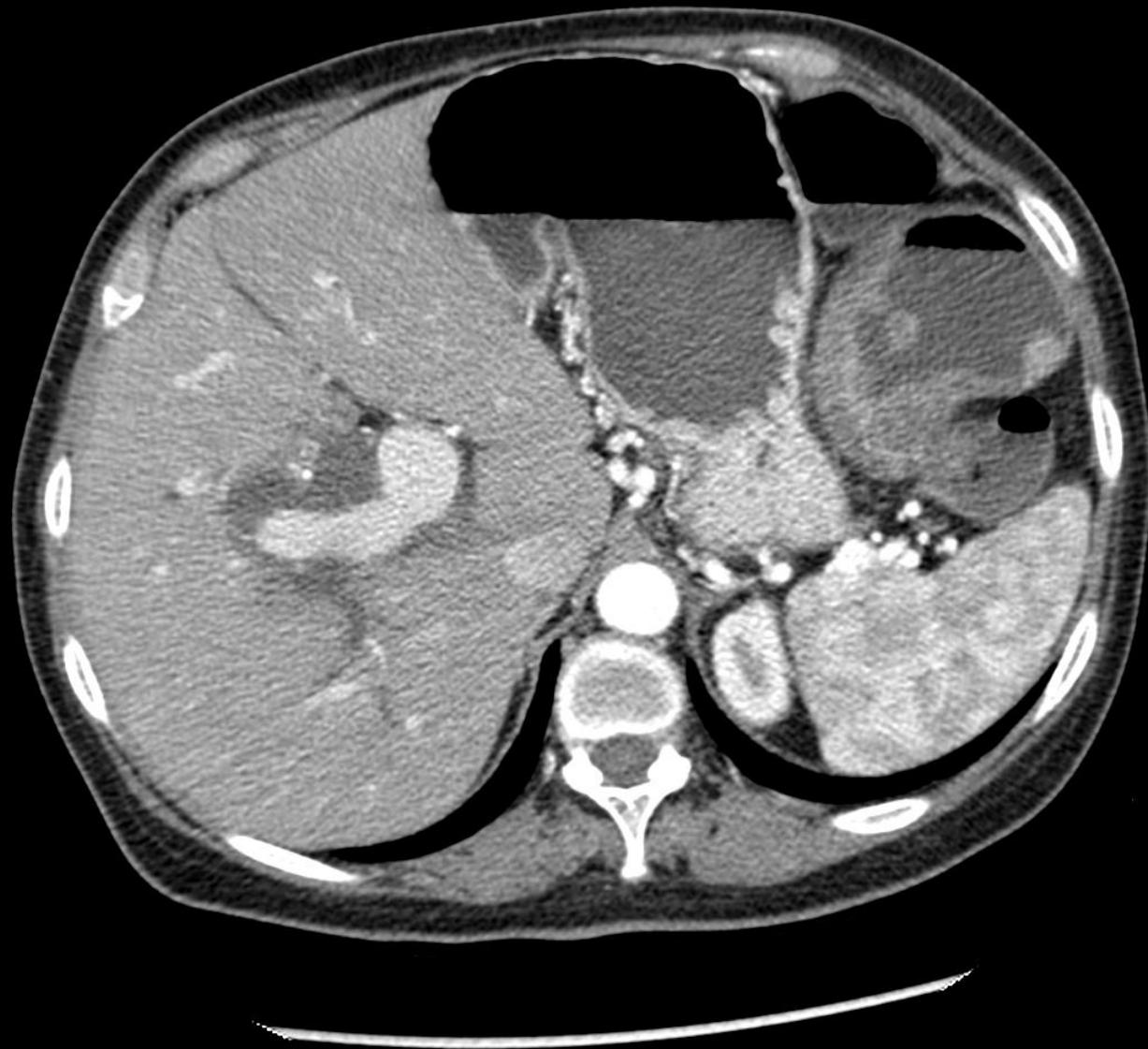
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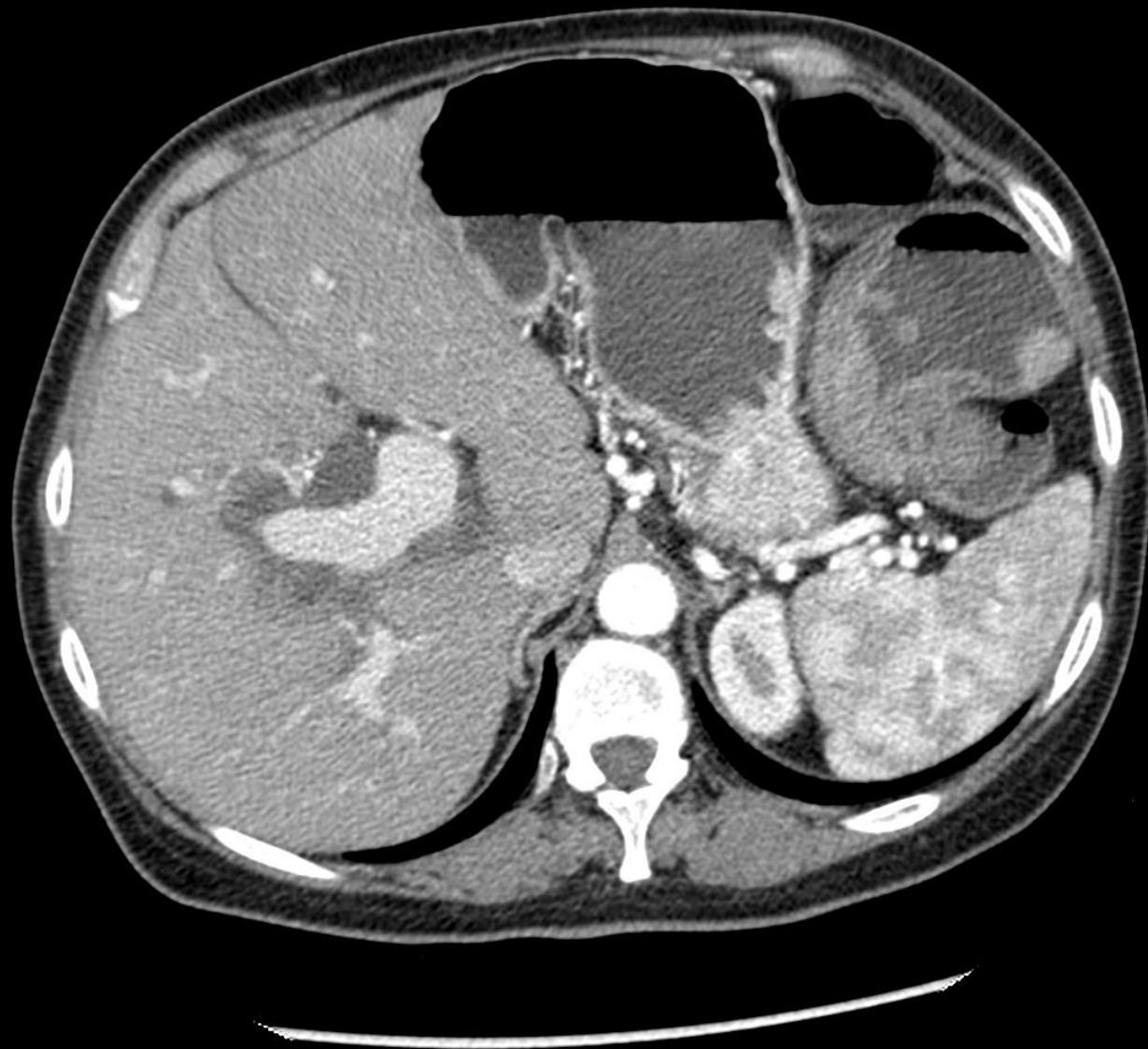
TC abdominal

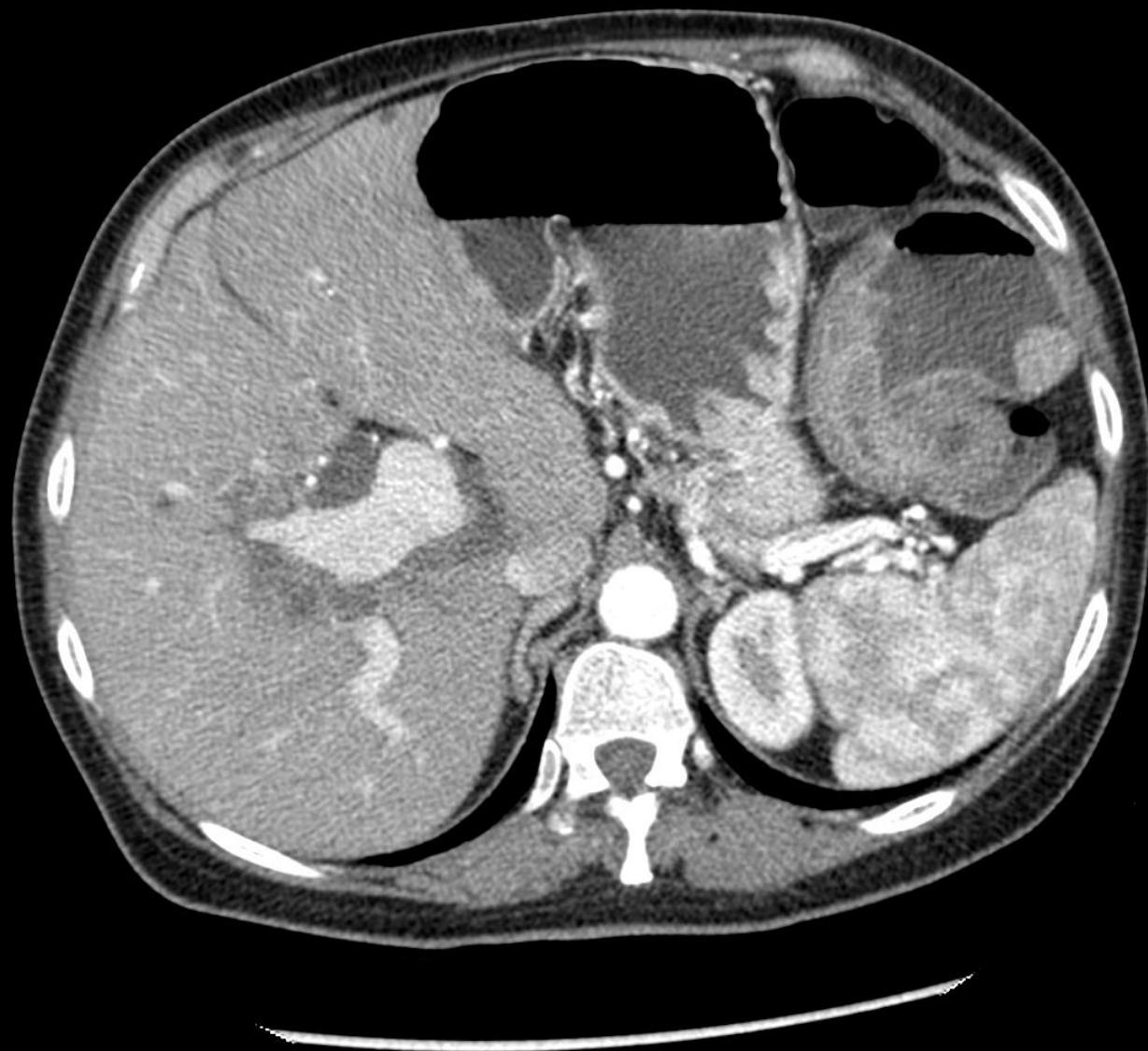






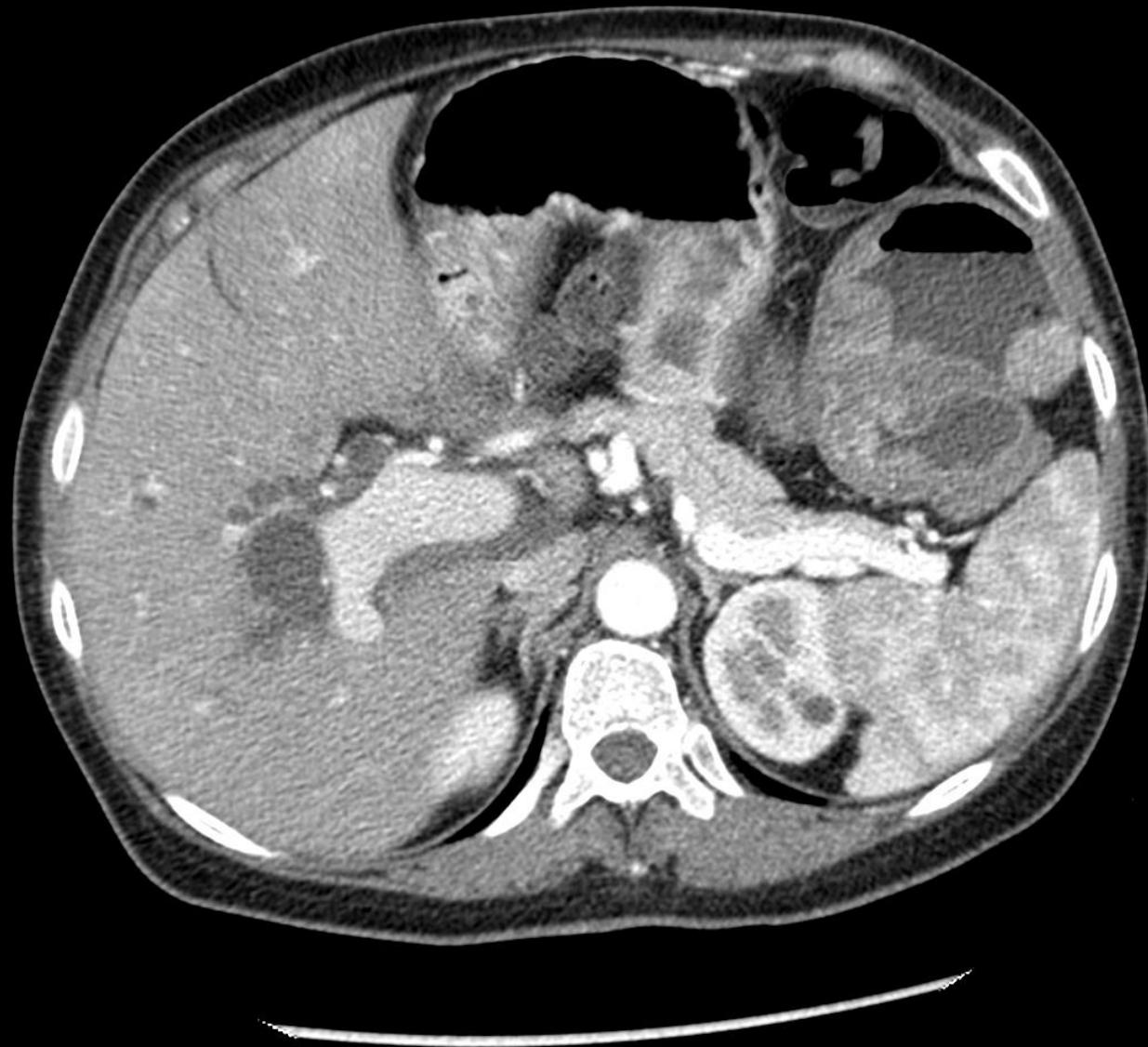


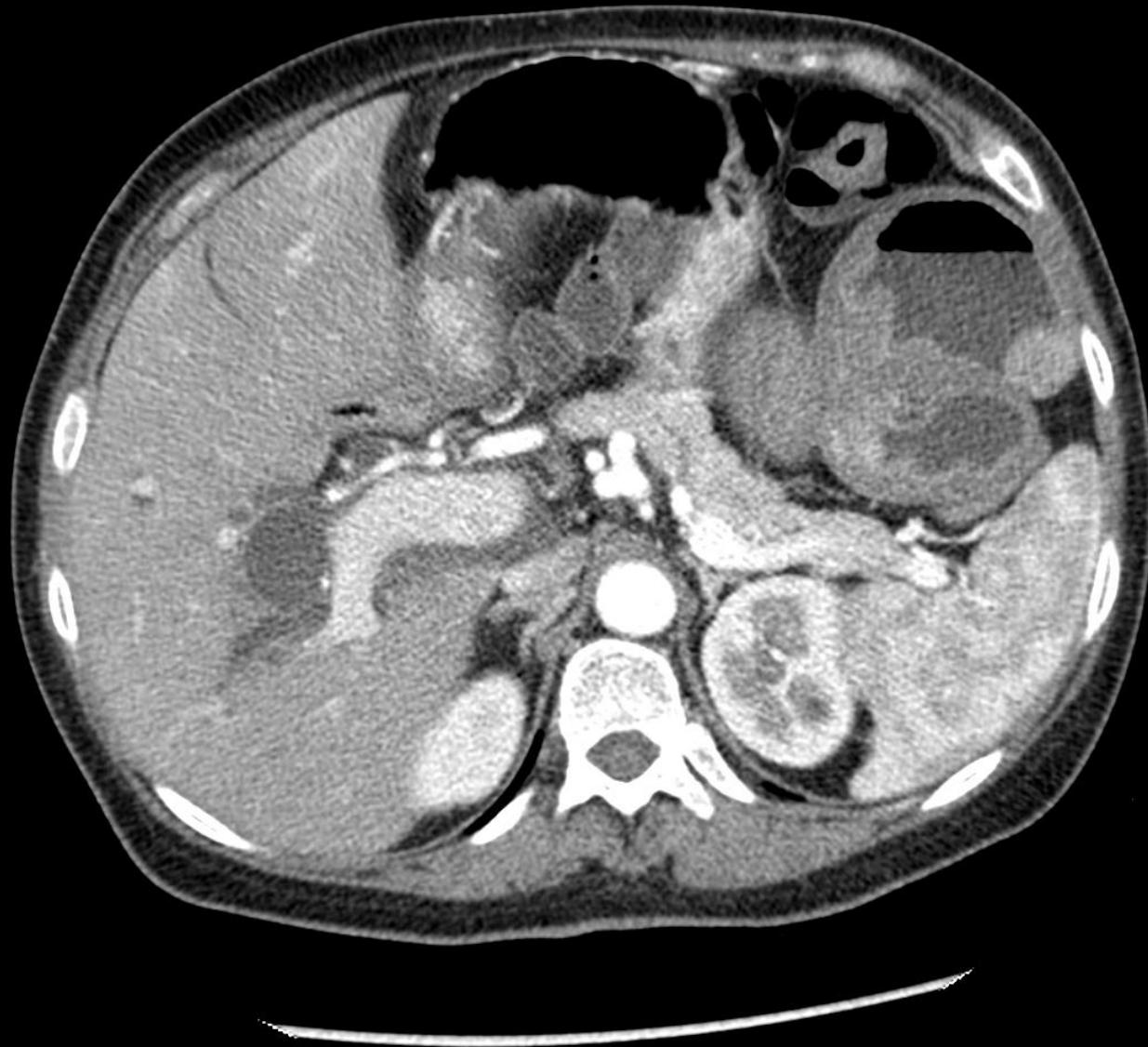


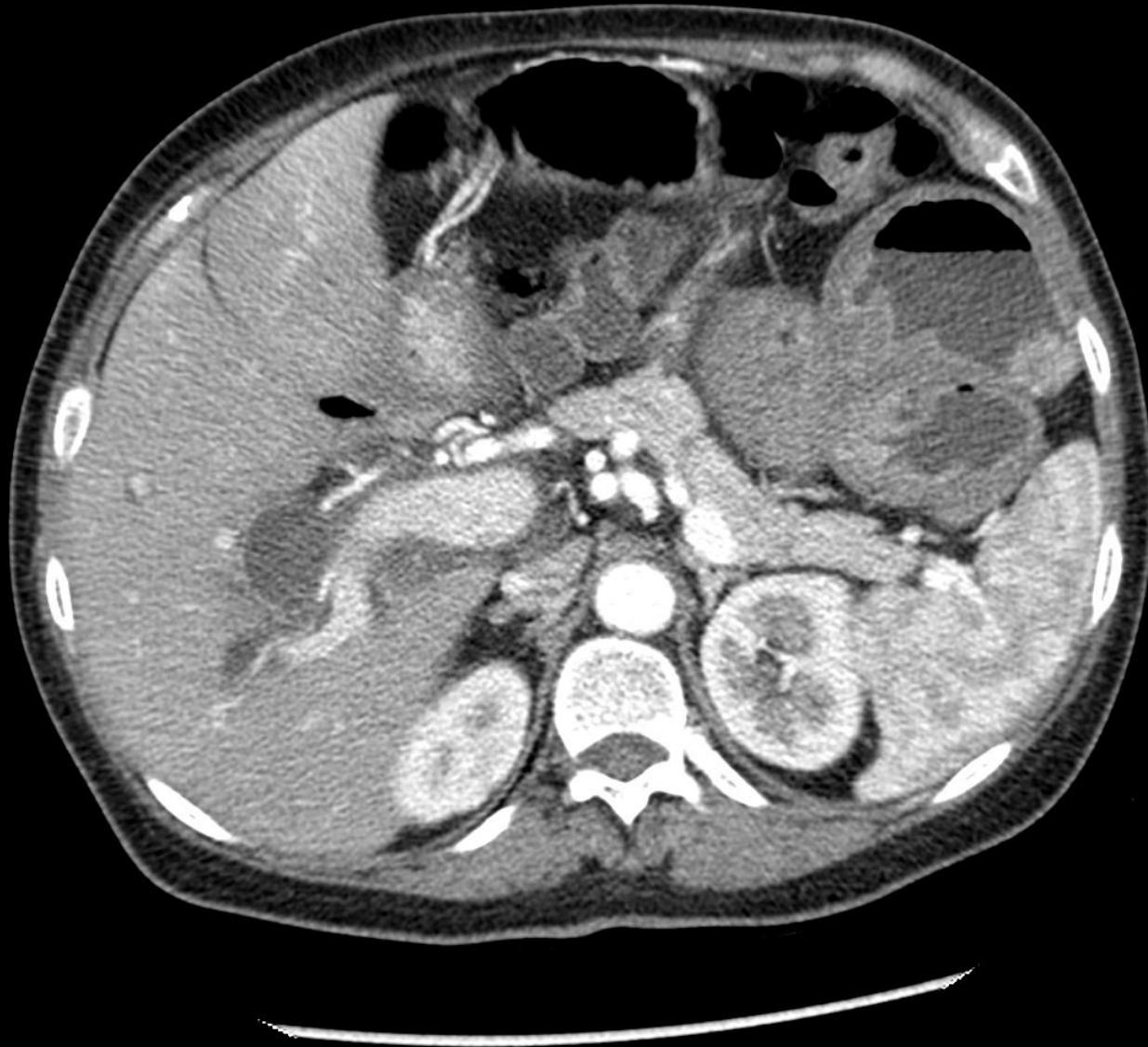


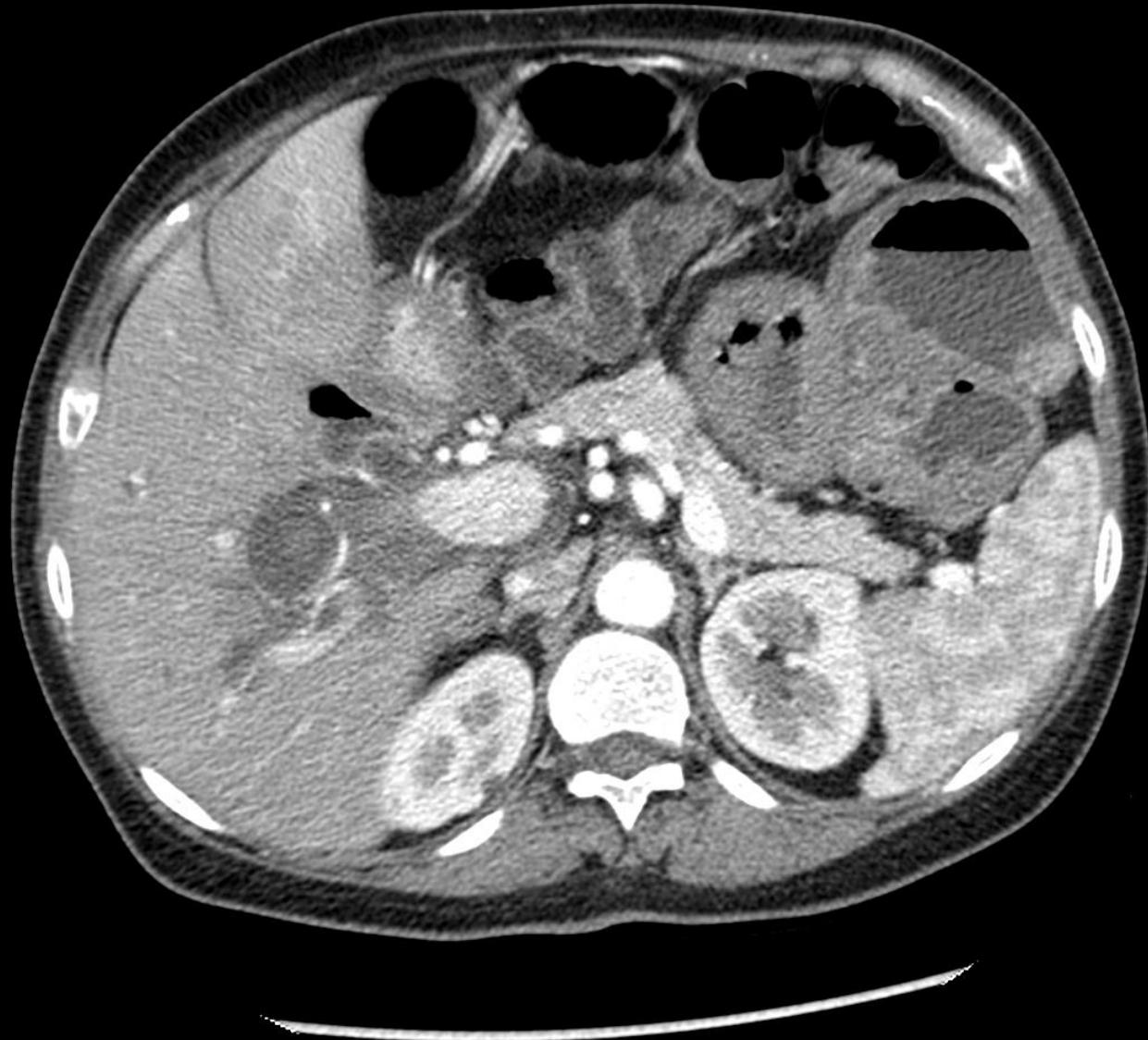






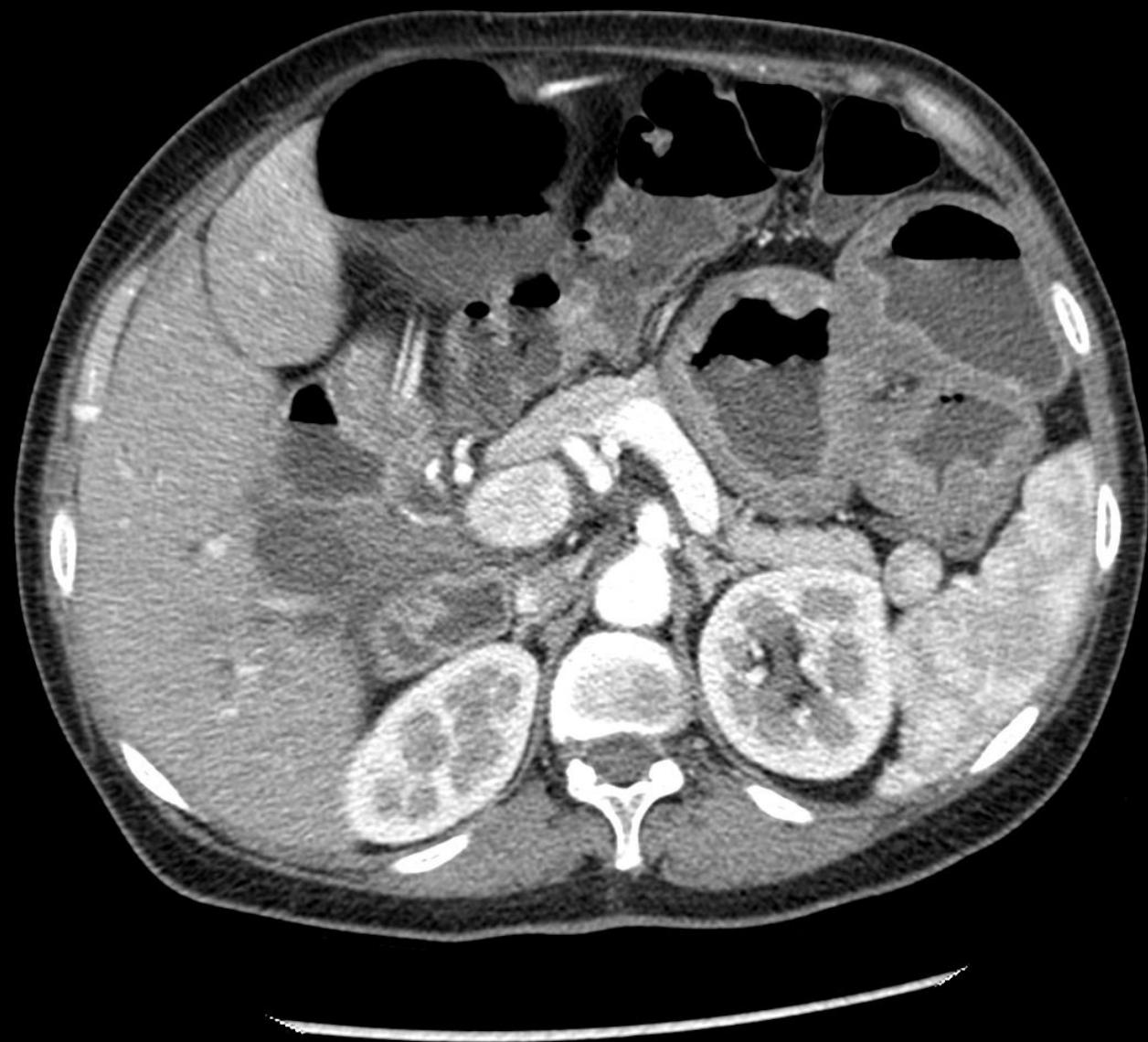


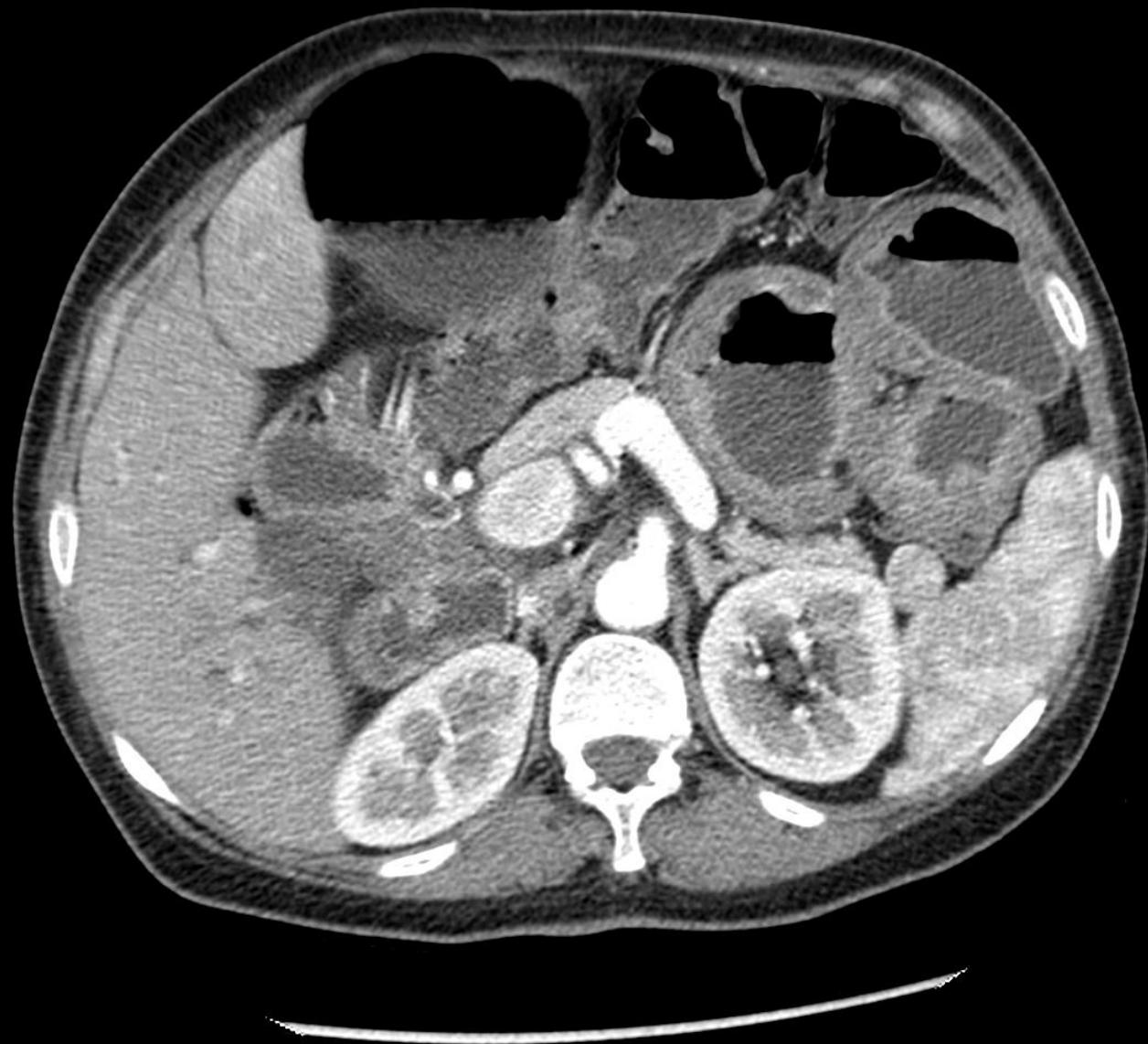


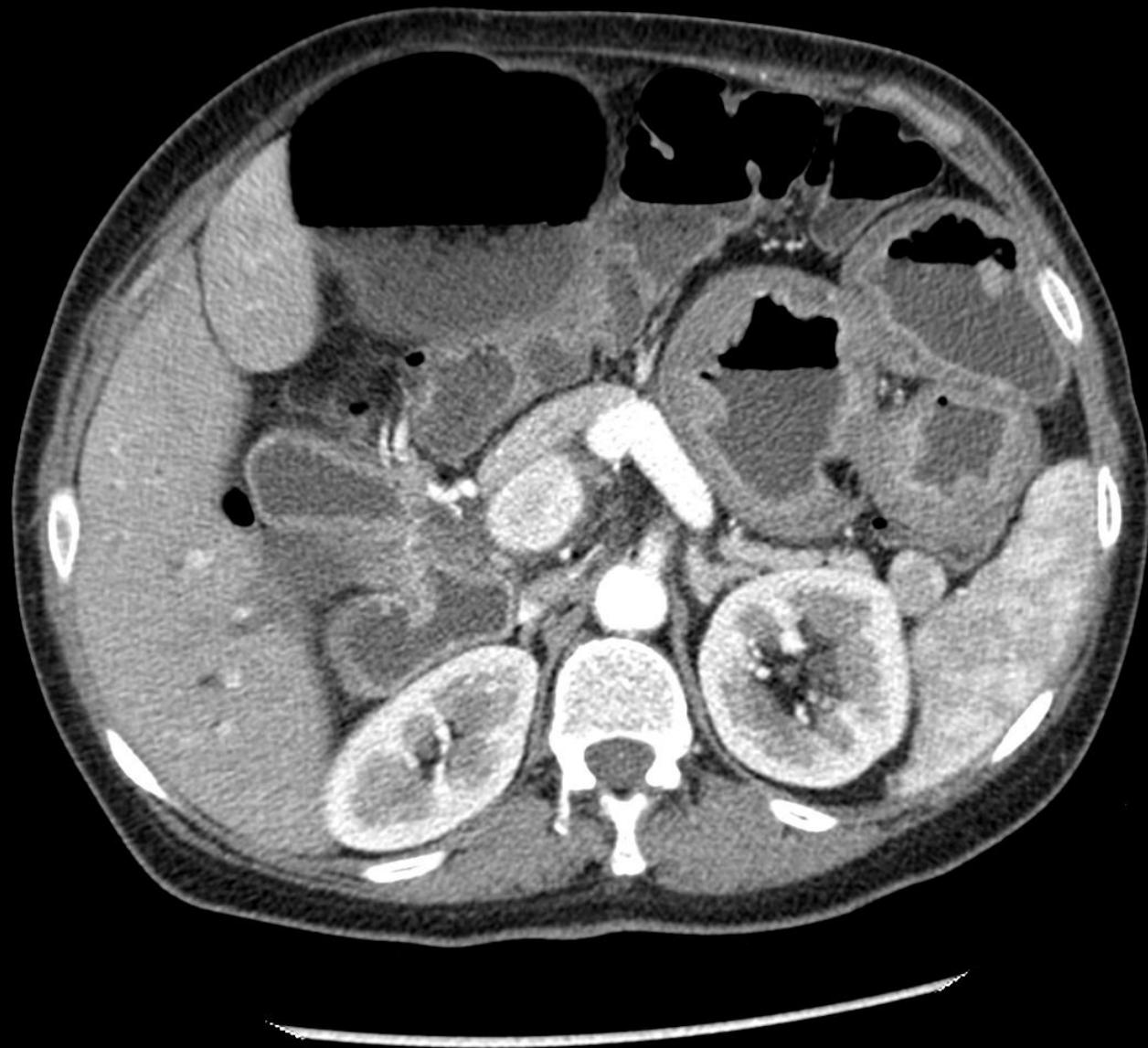


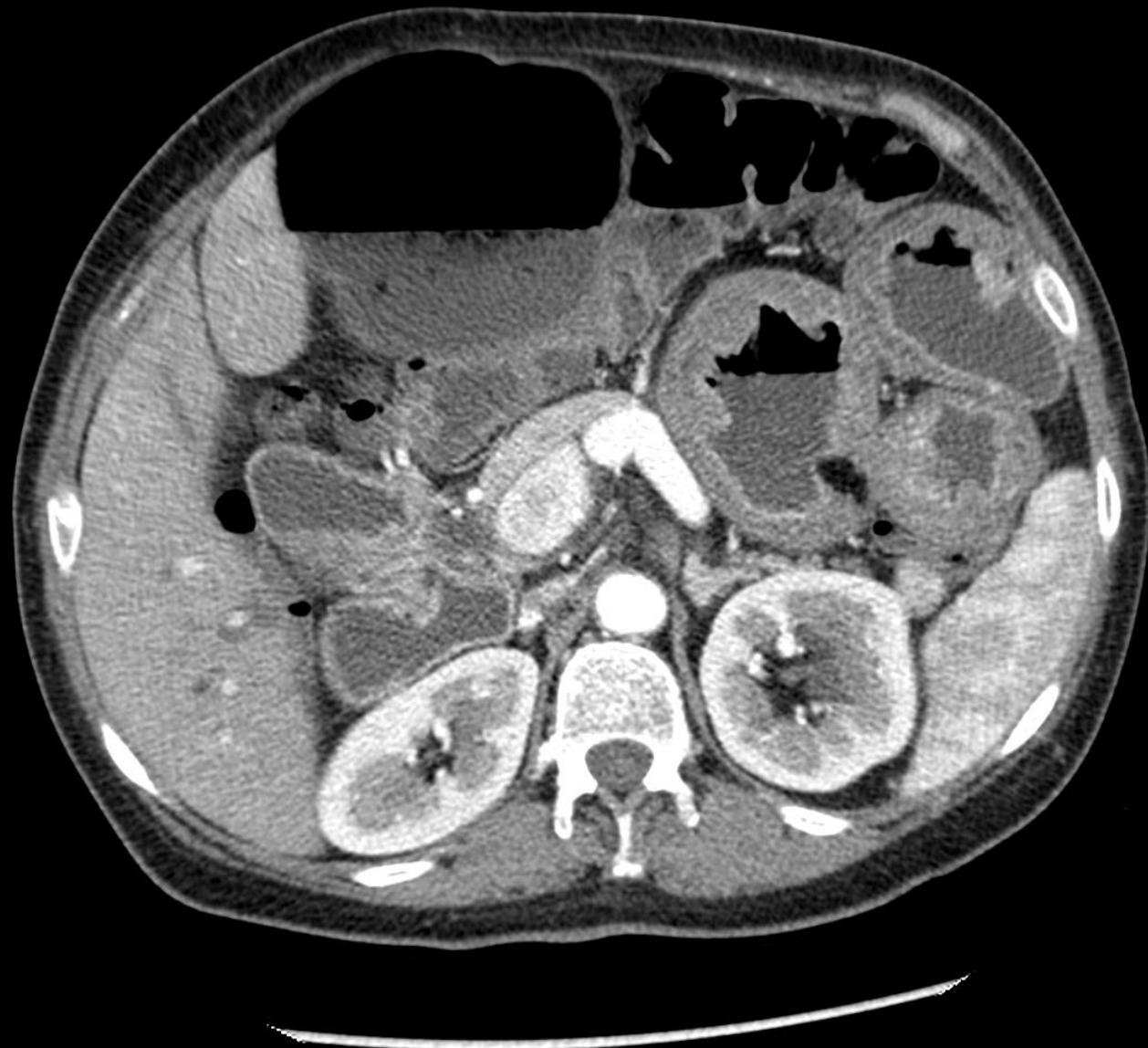


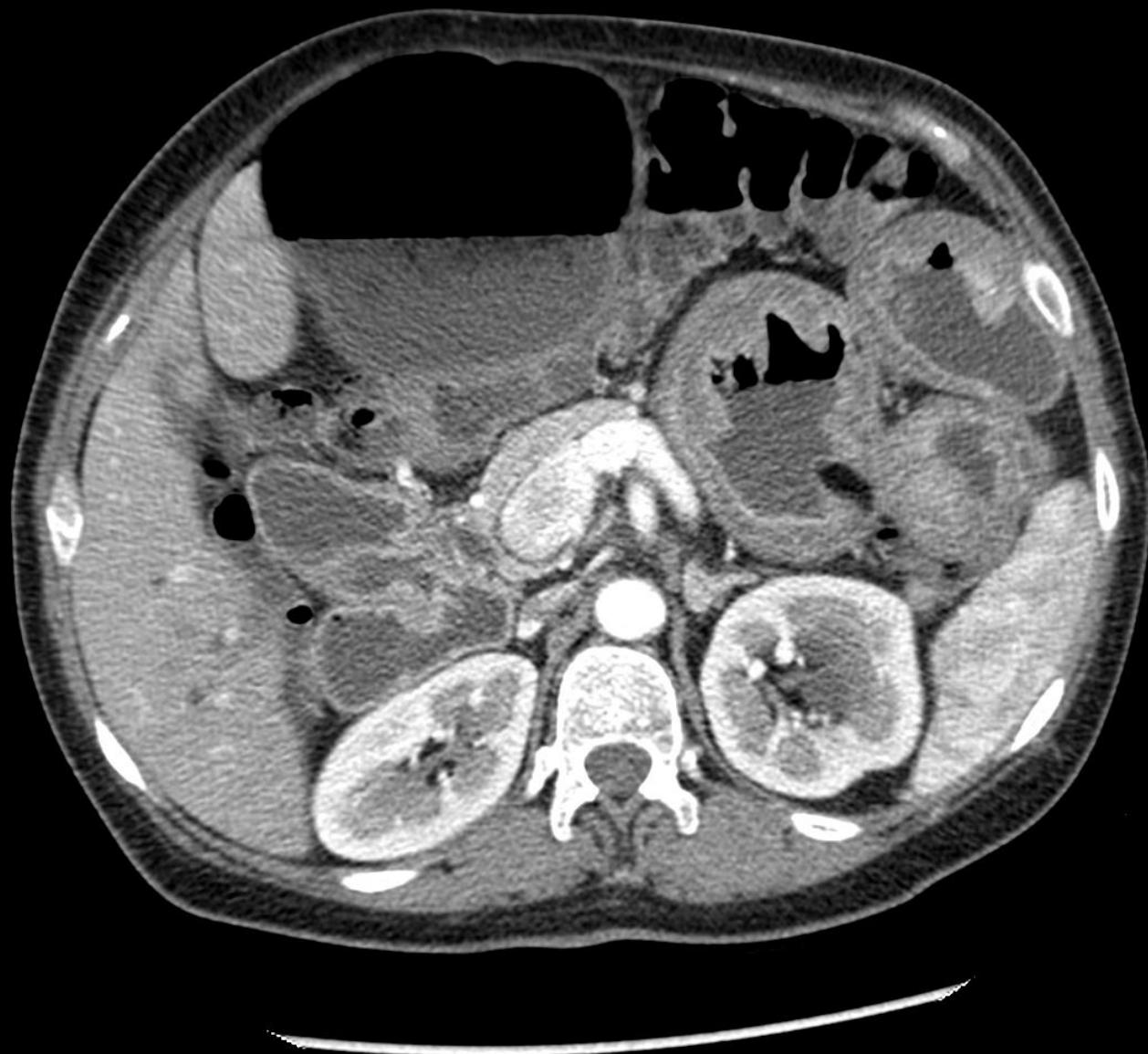


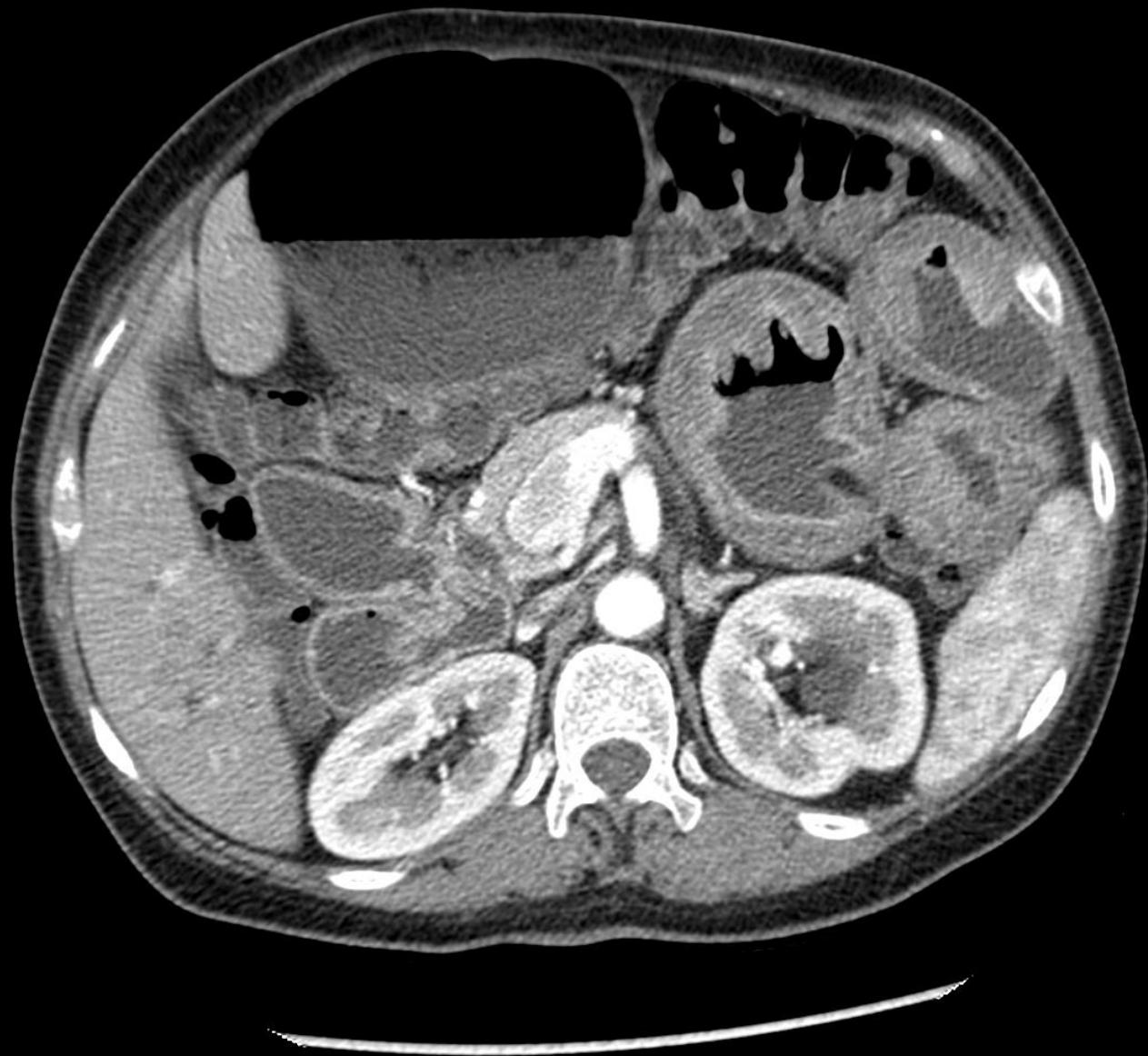


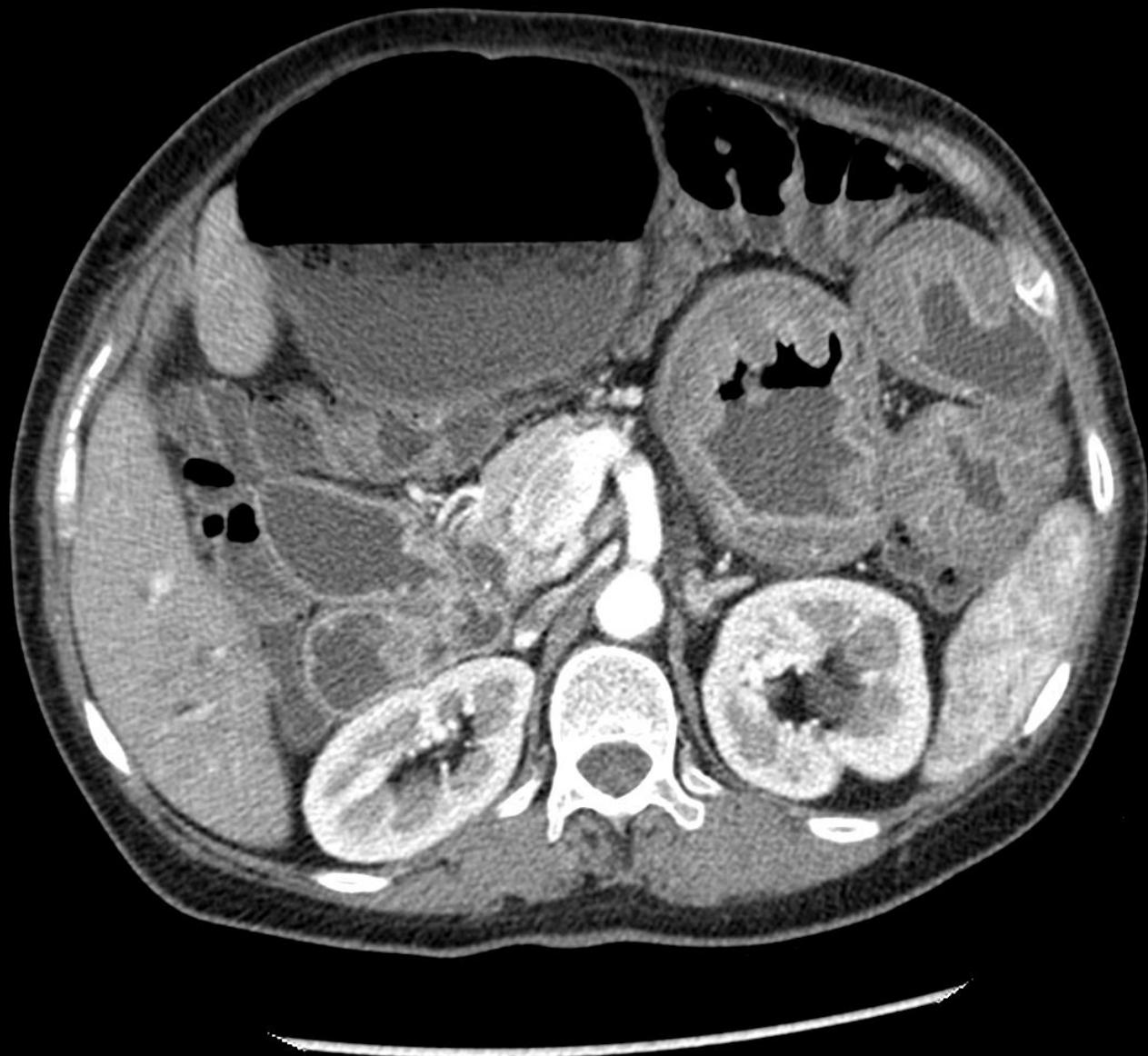


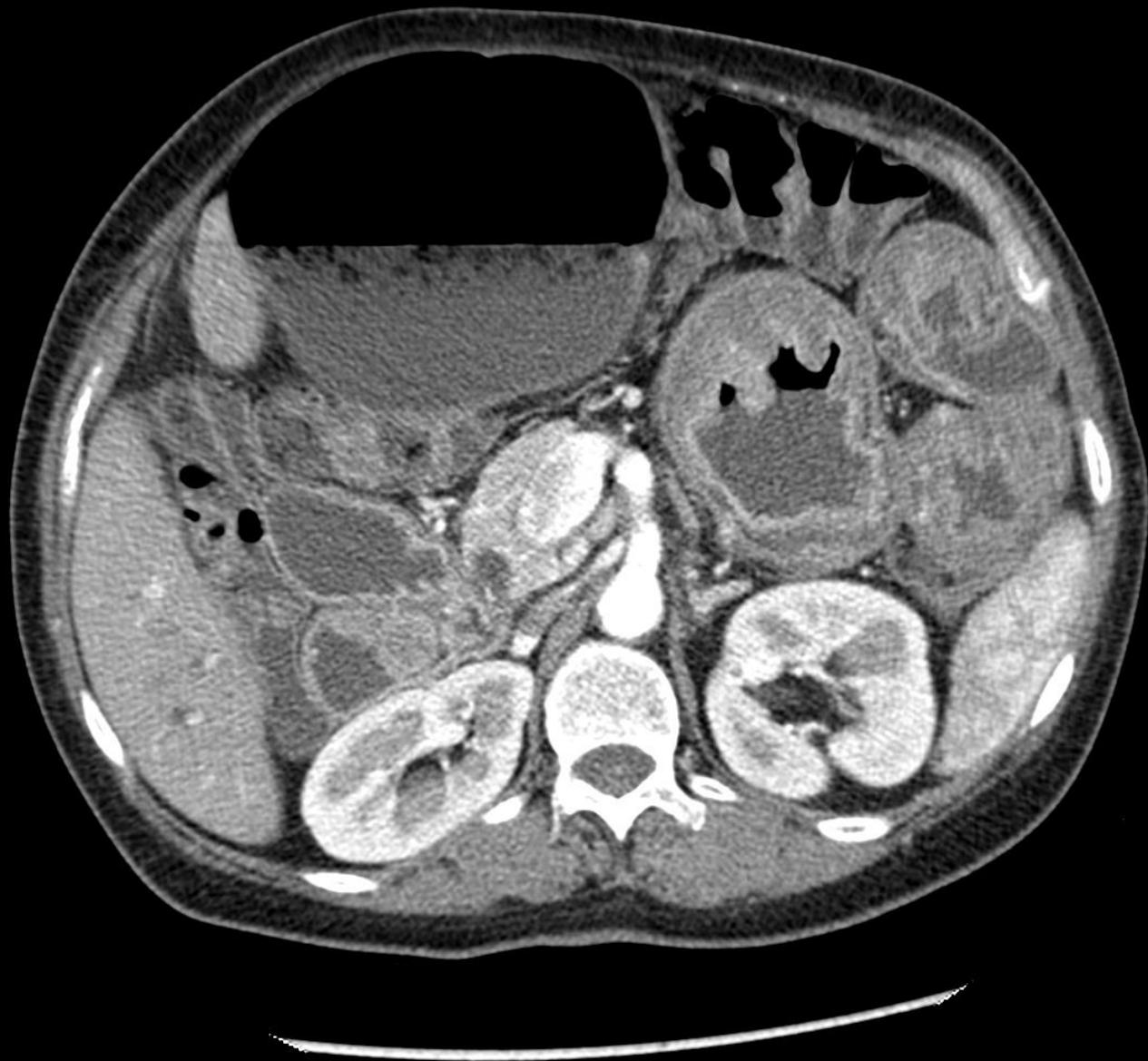


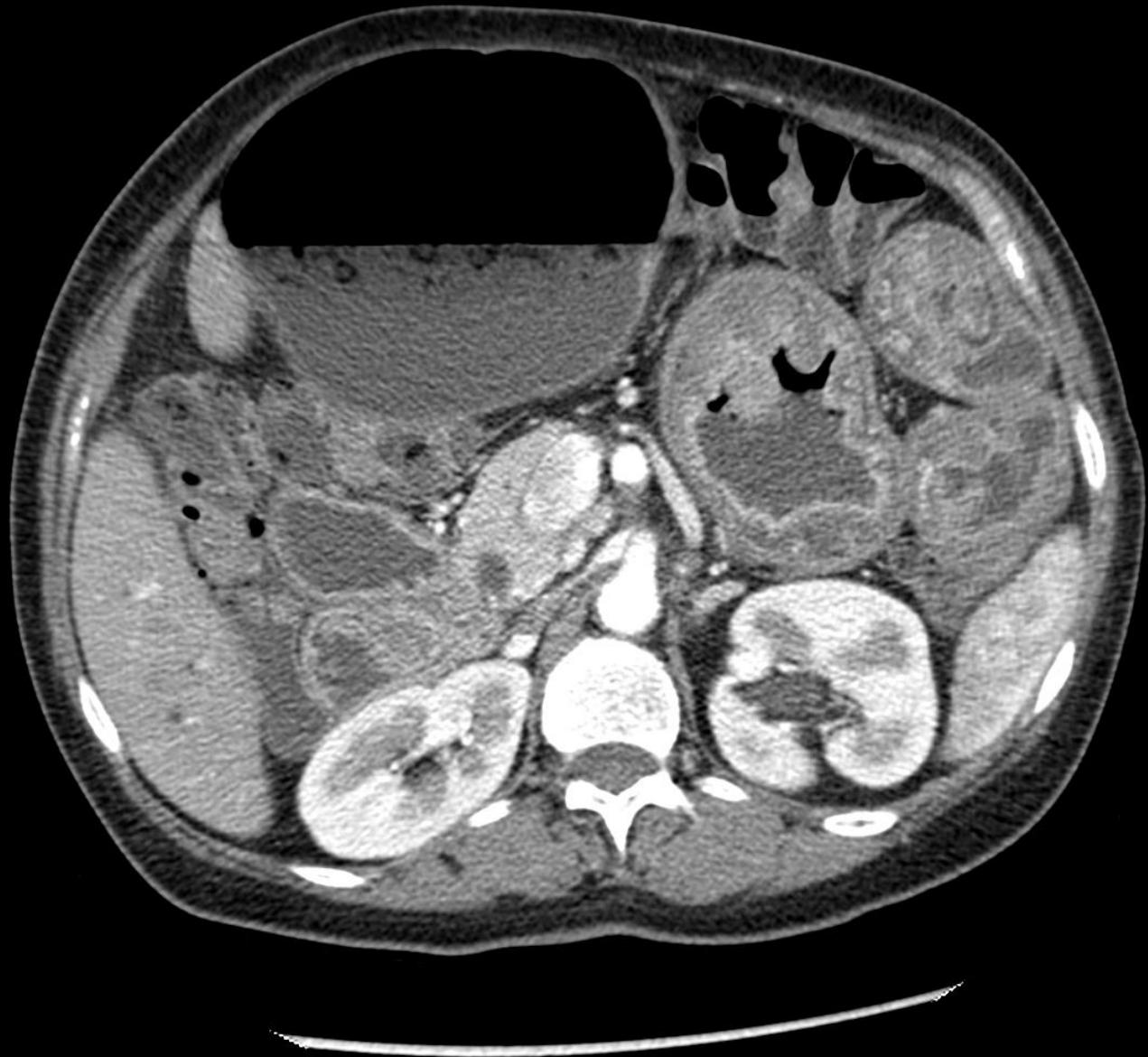


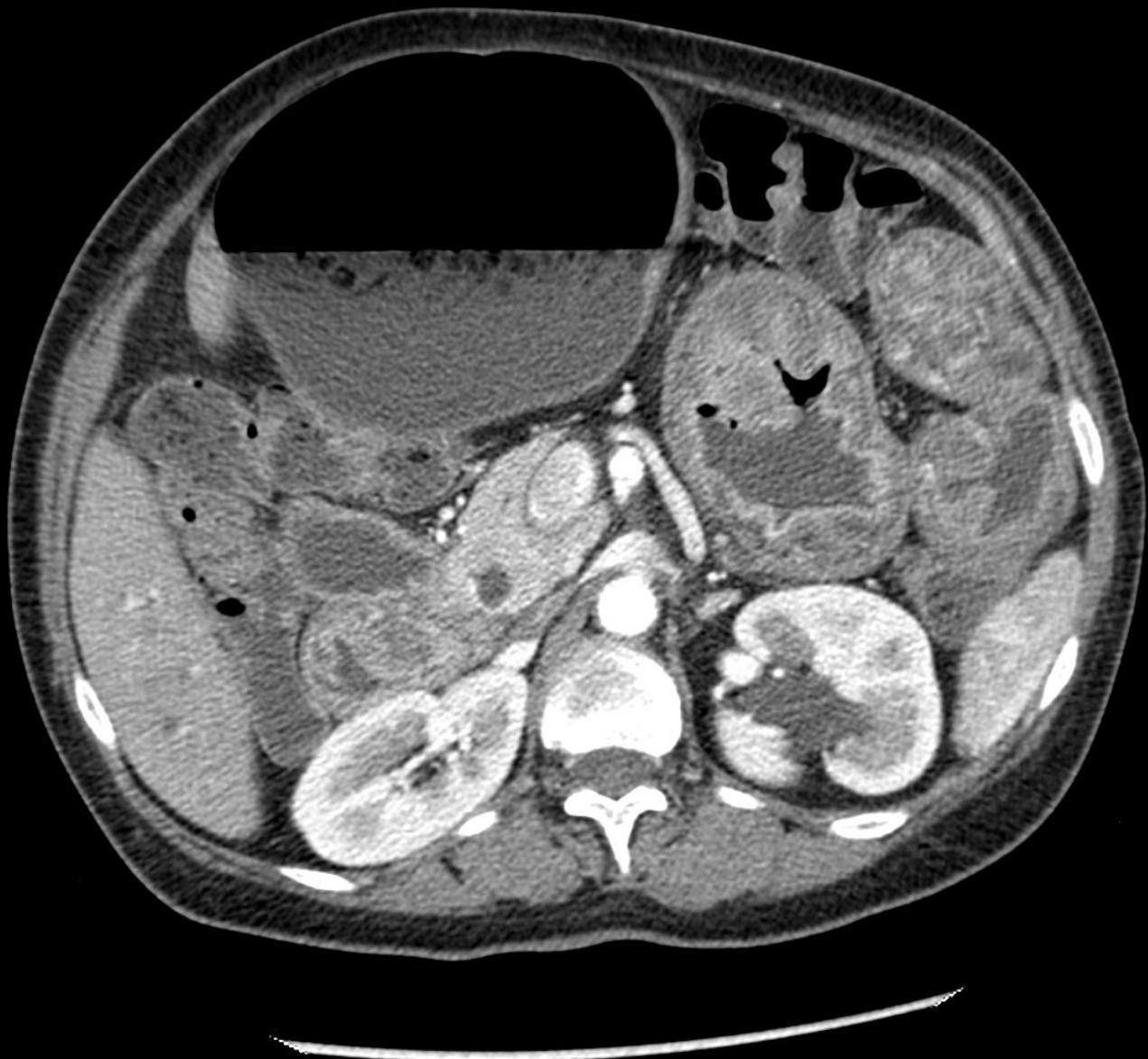








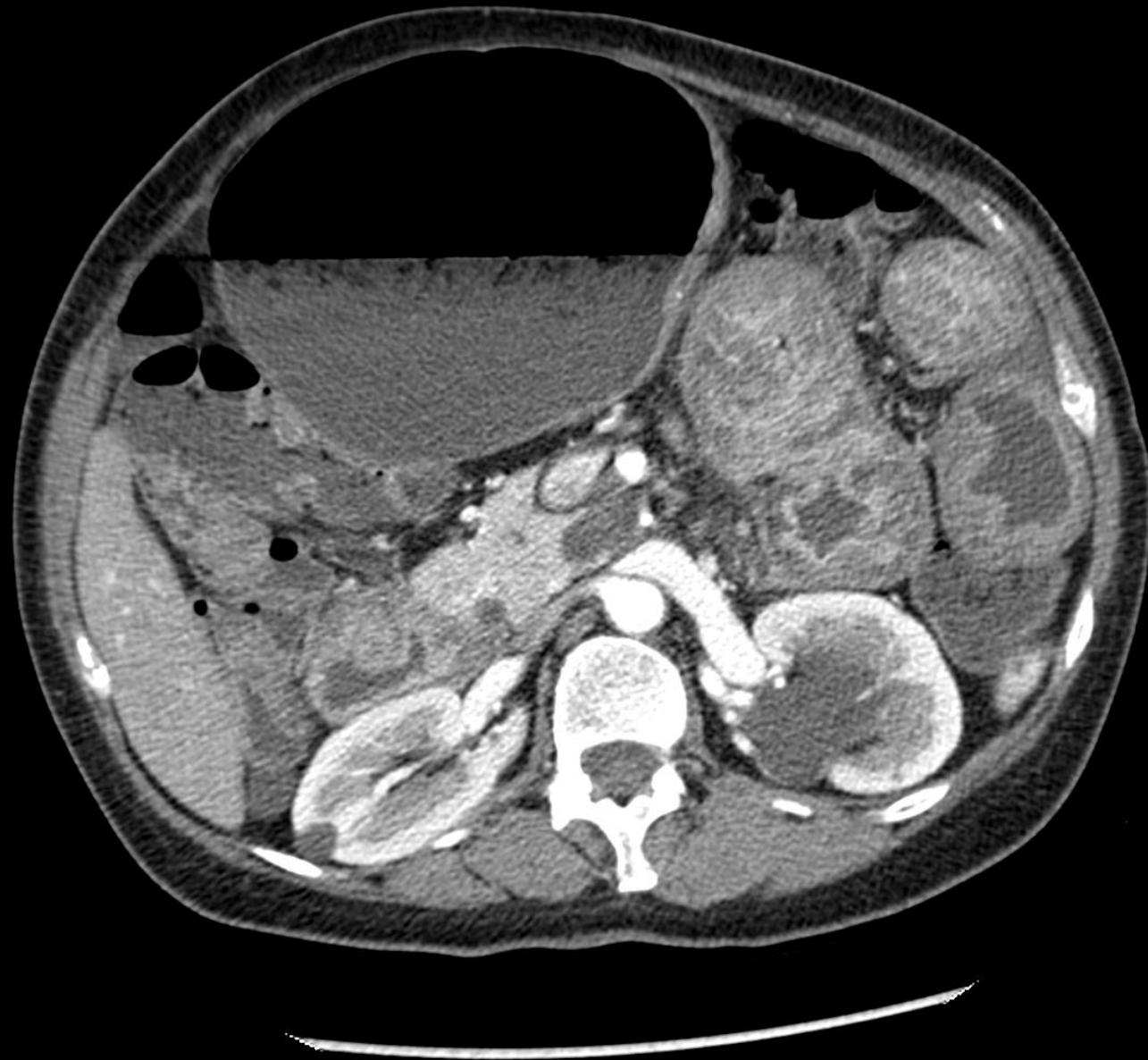


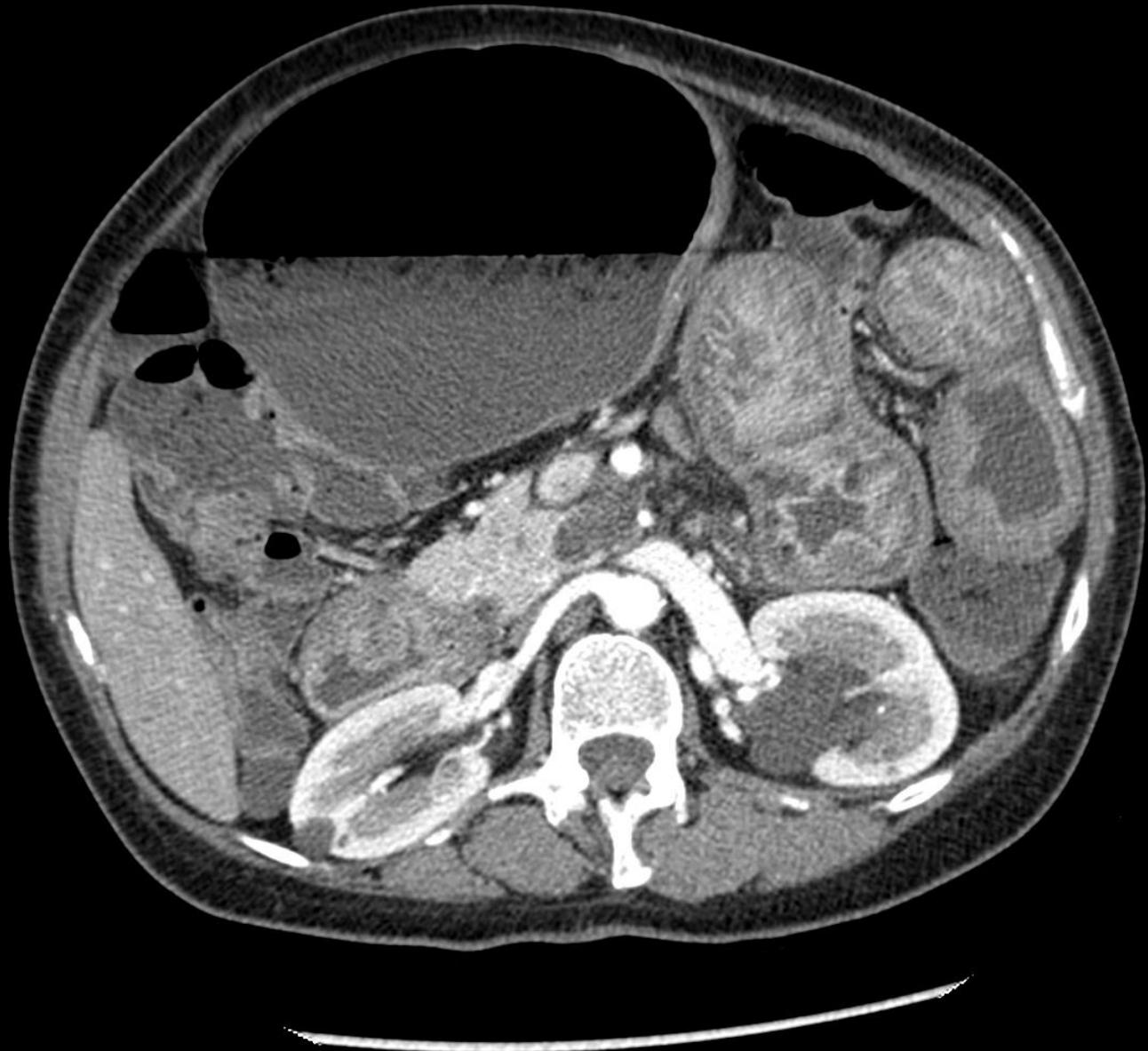


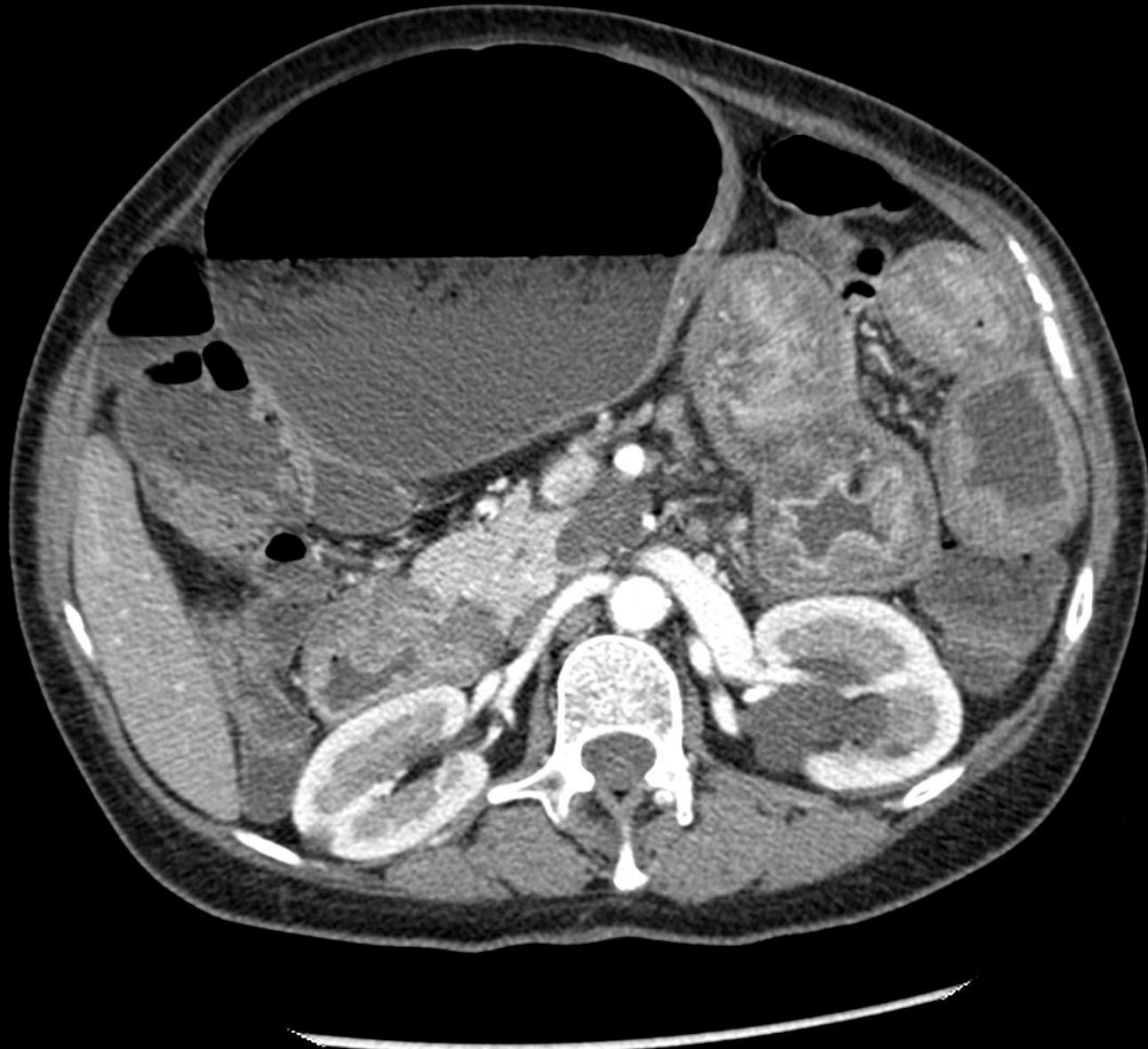










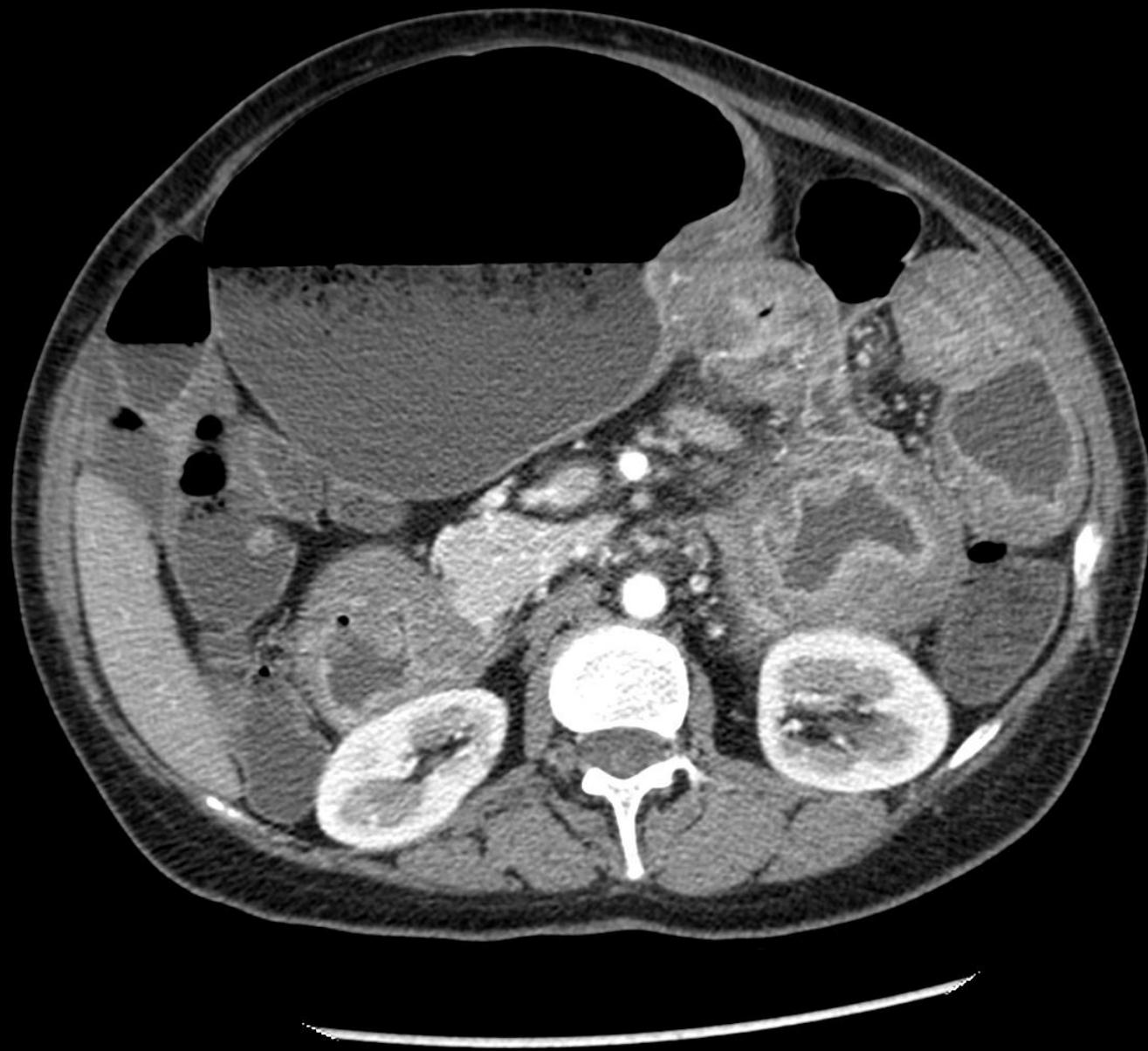


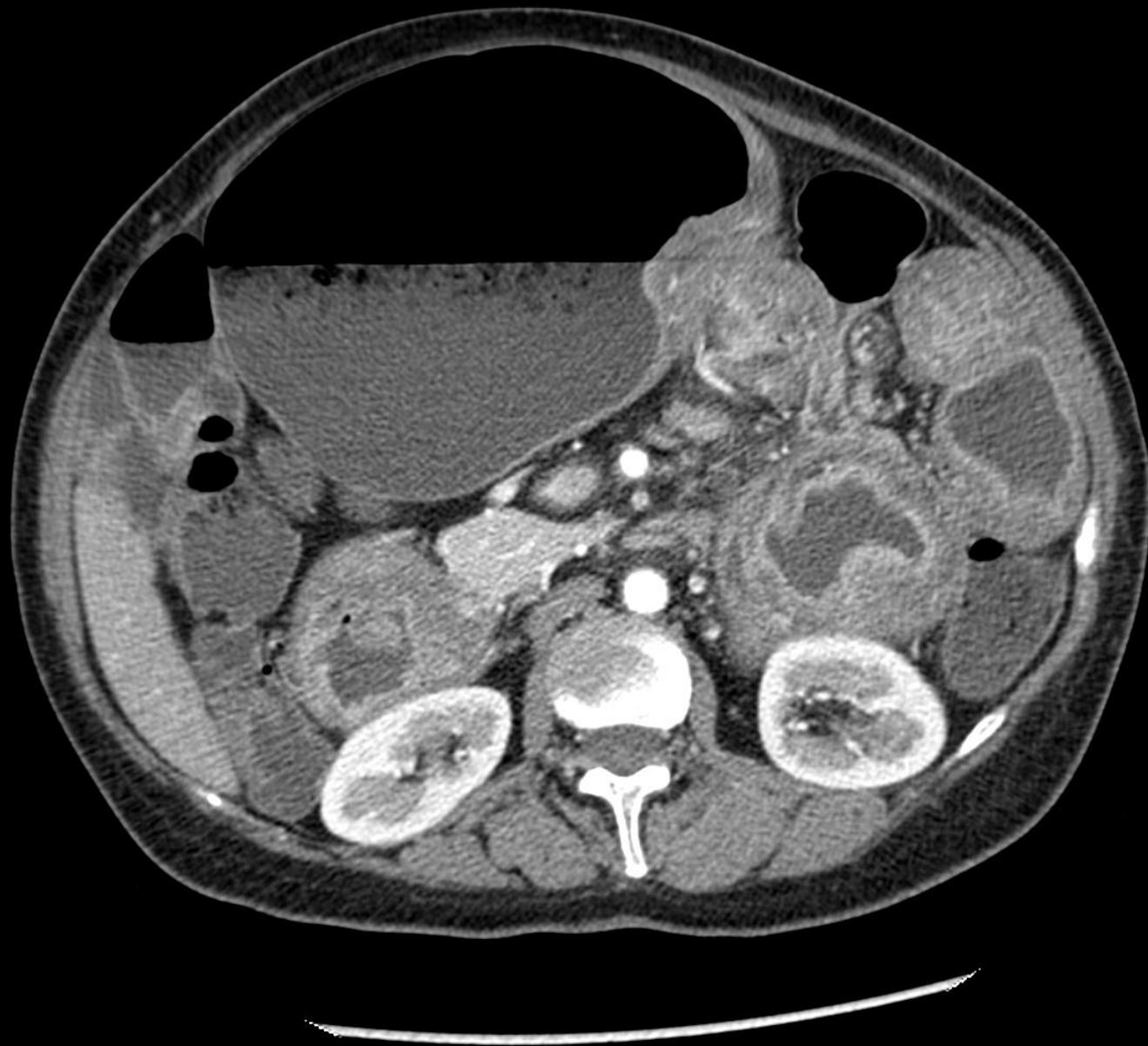




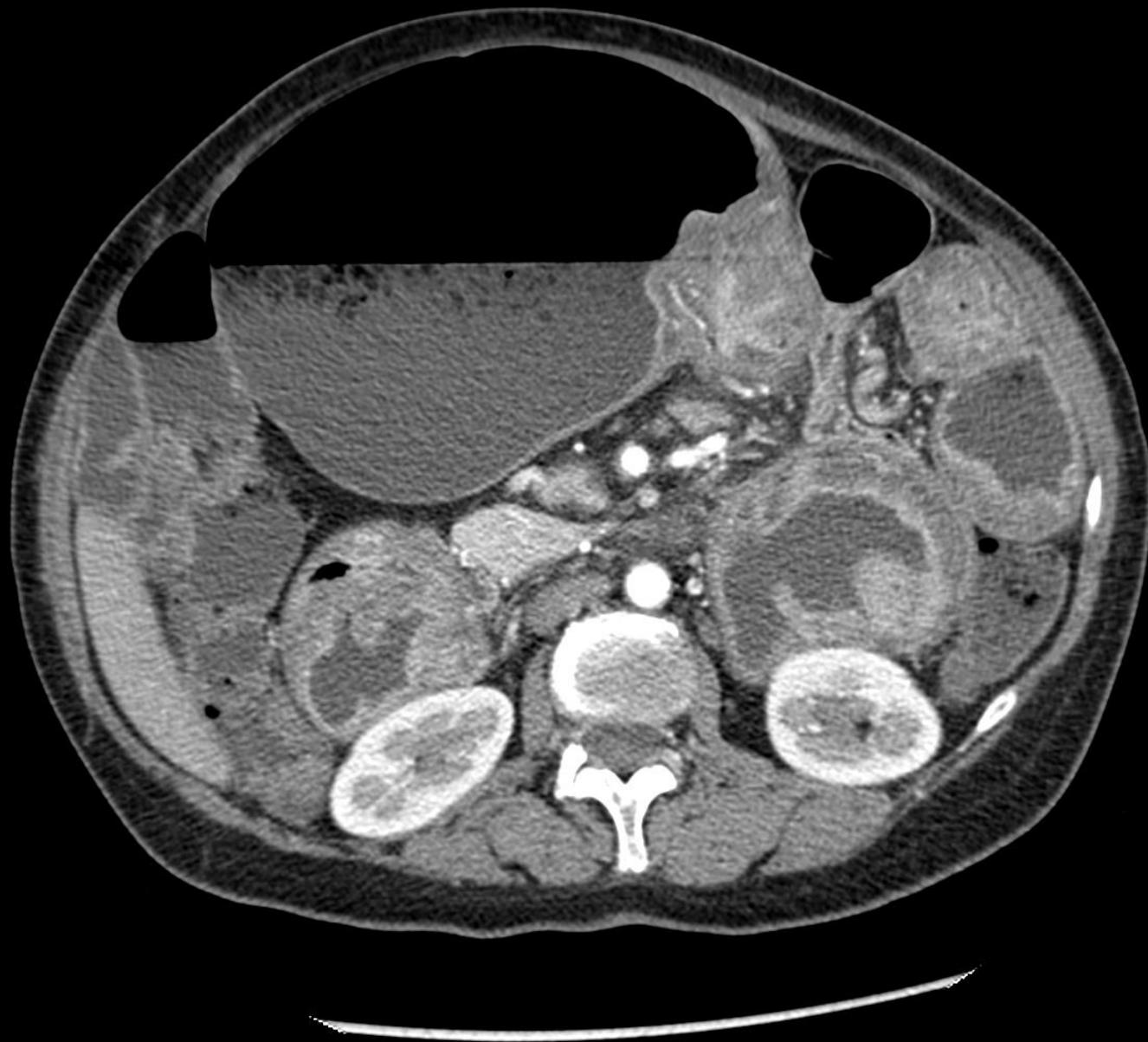


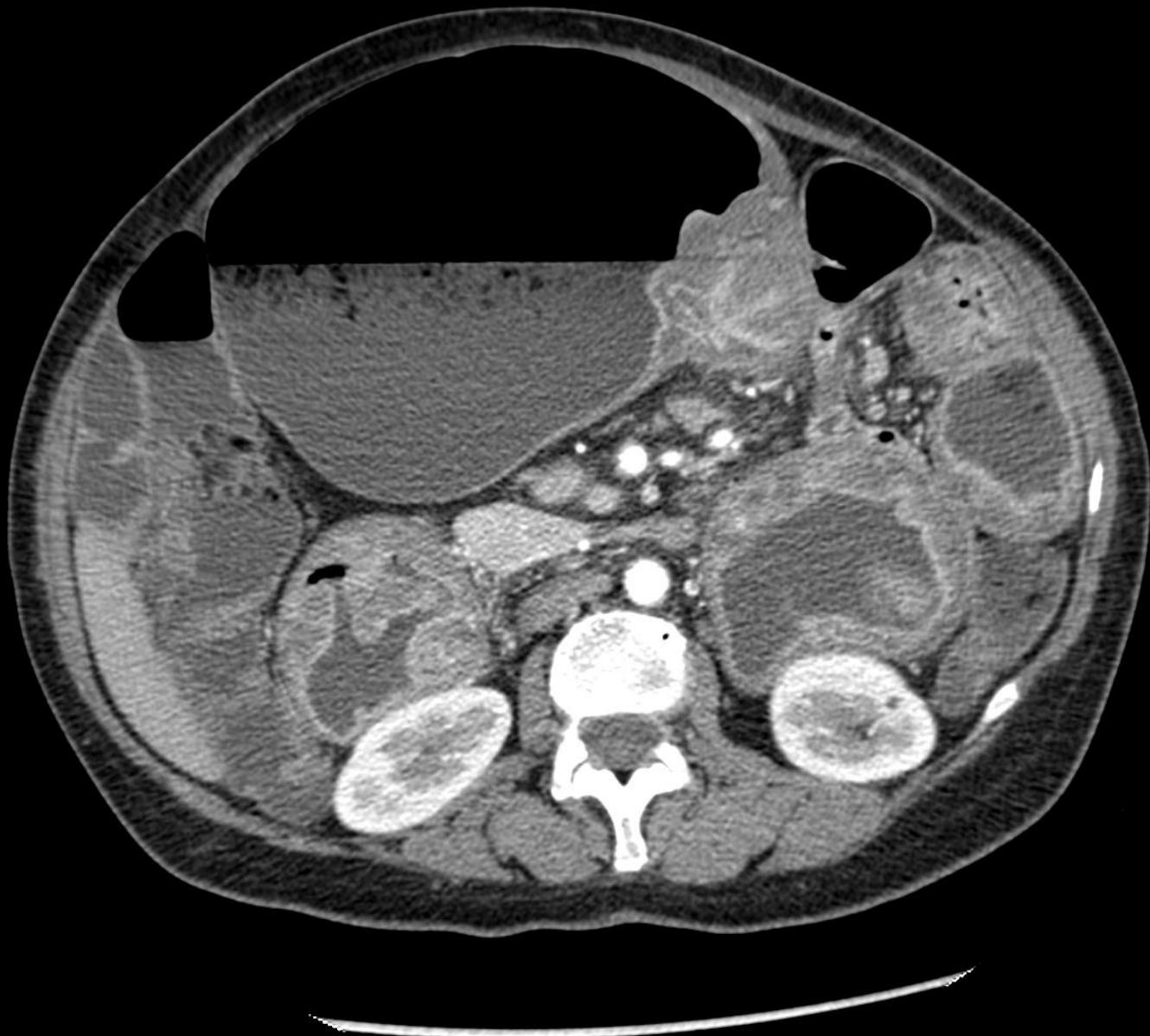


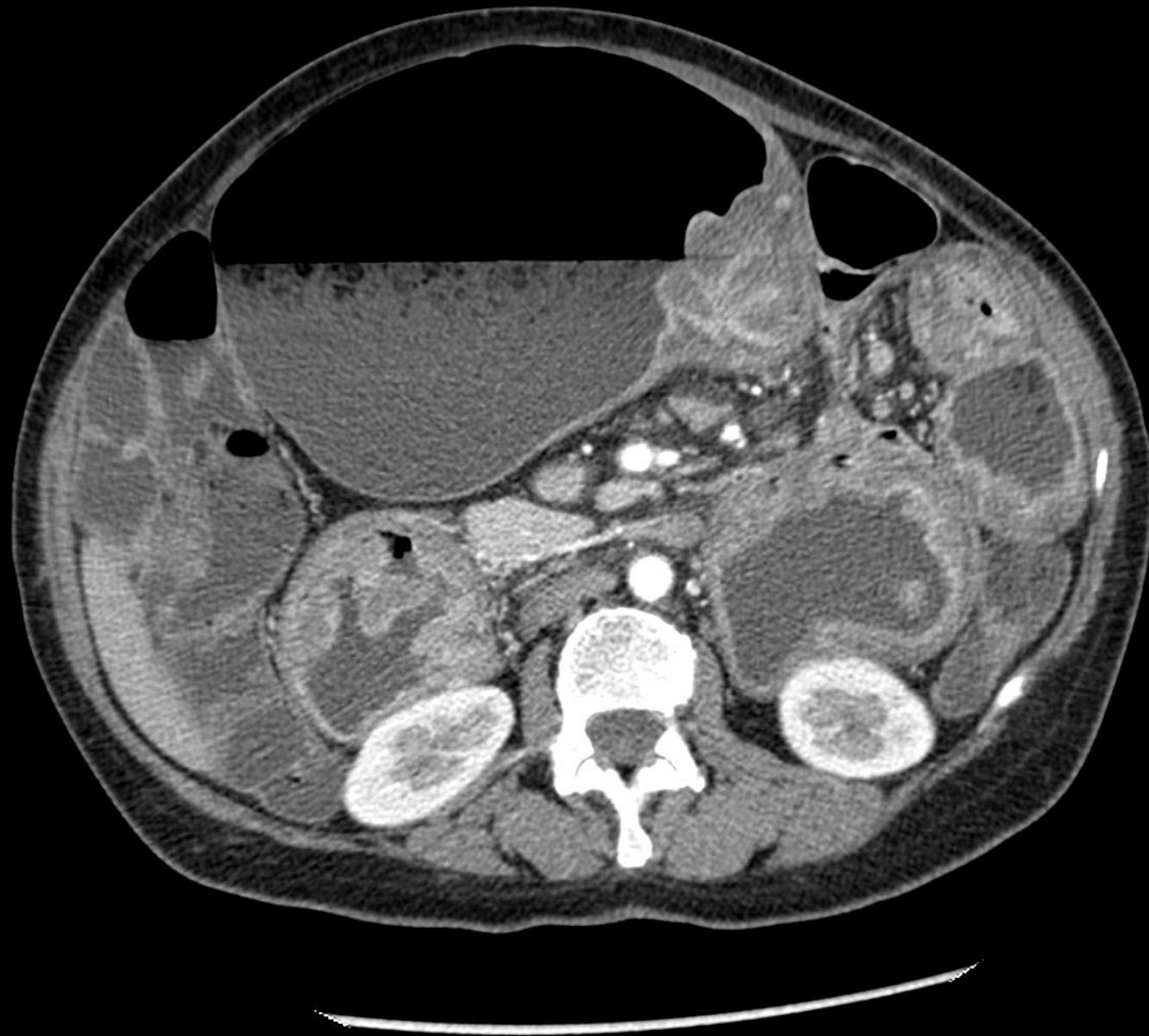


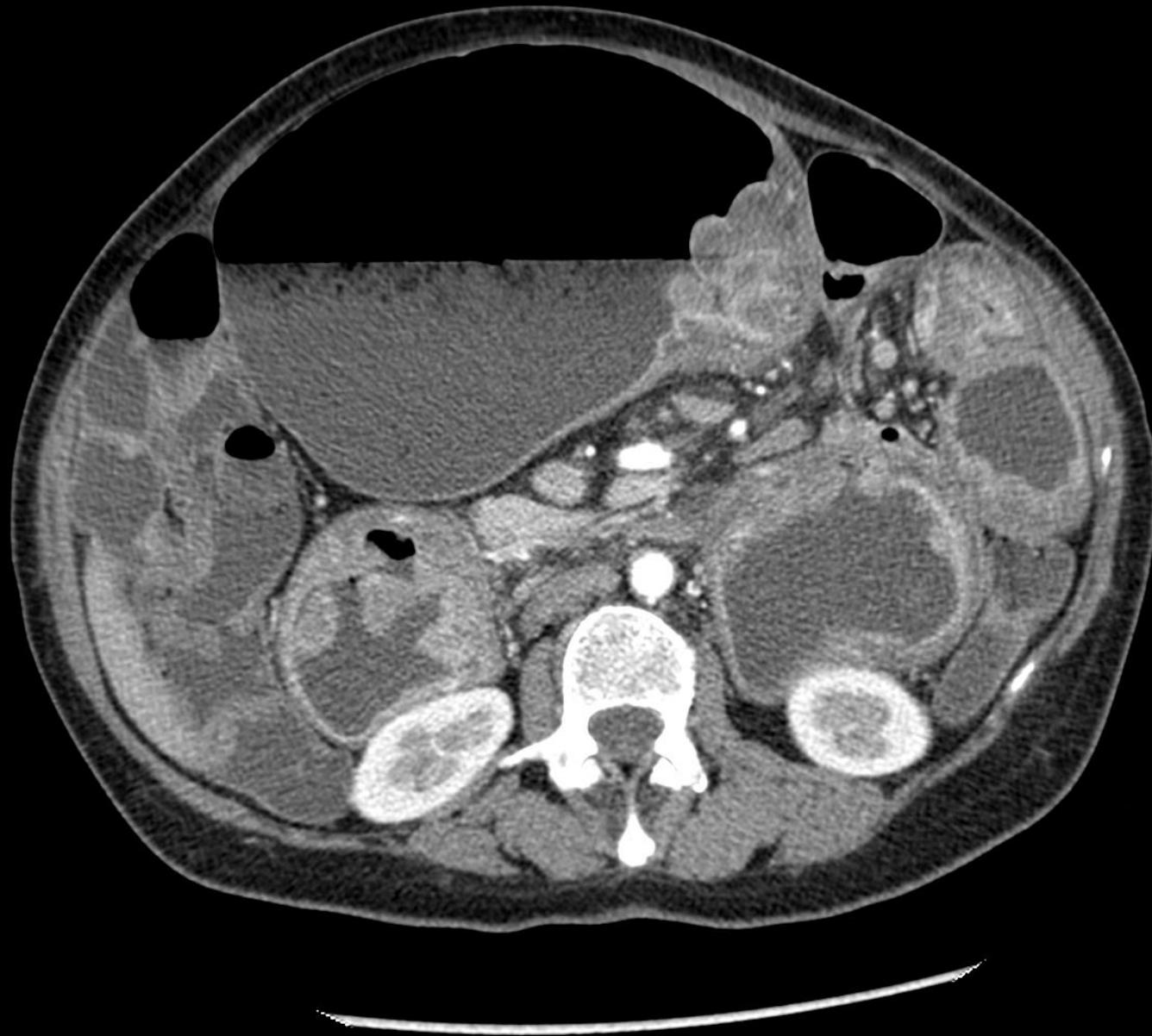




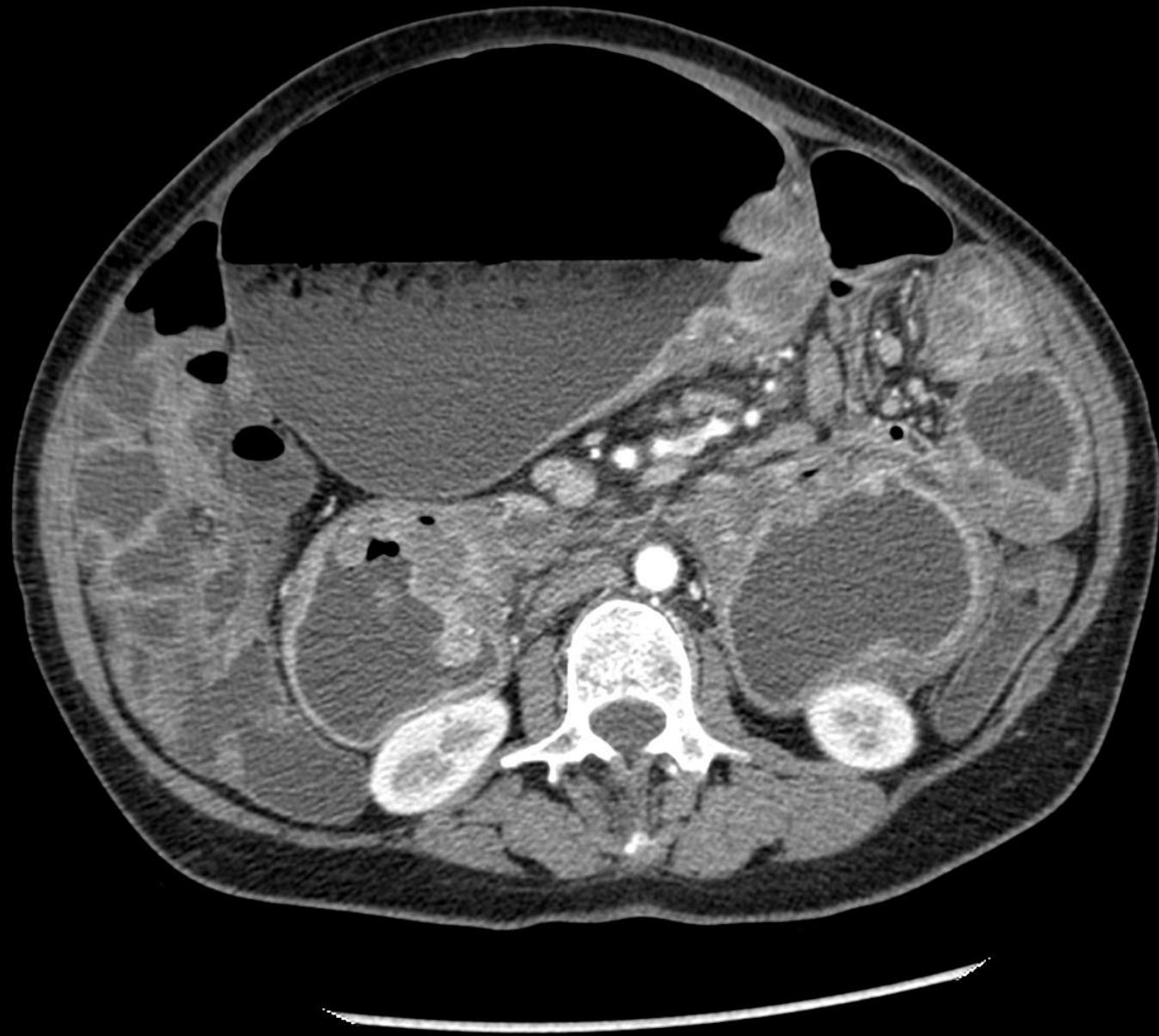


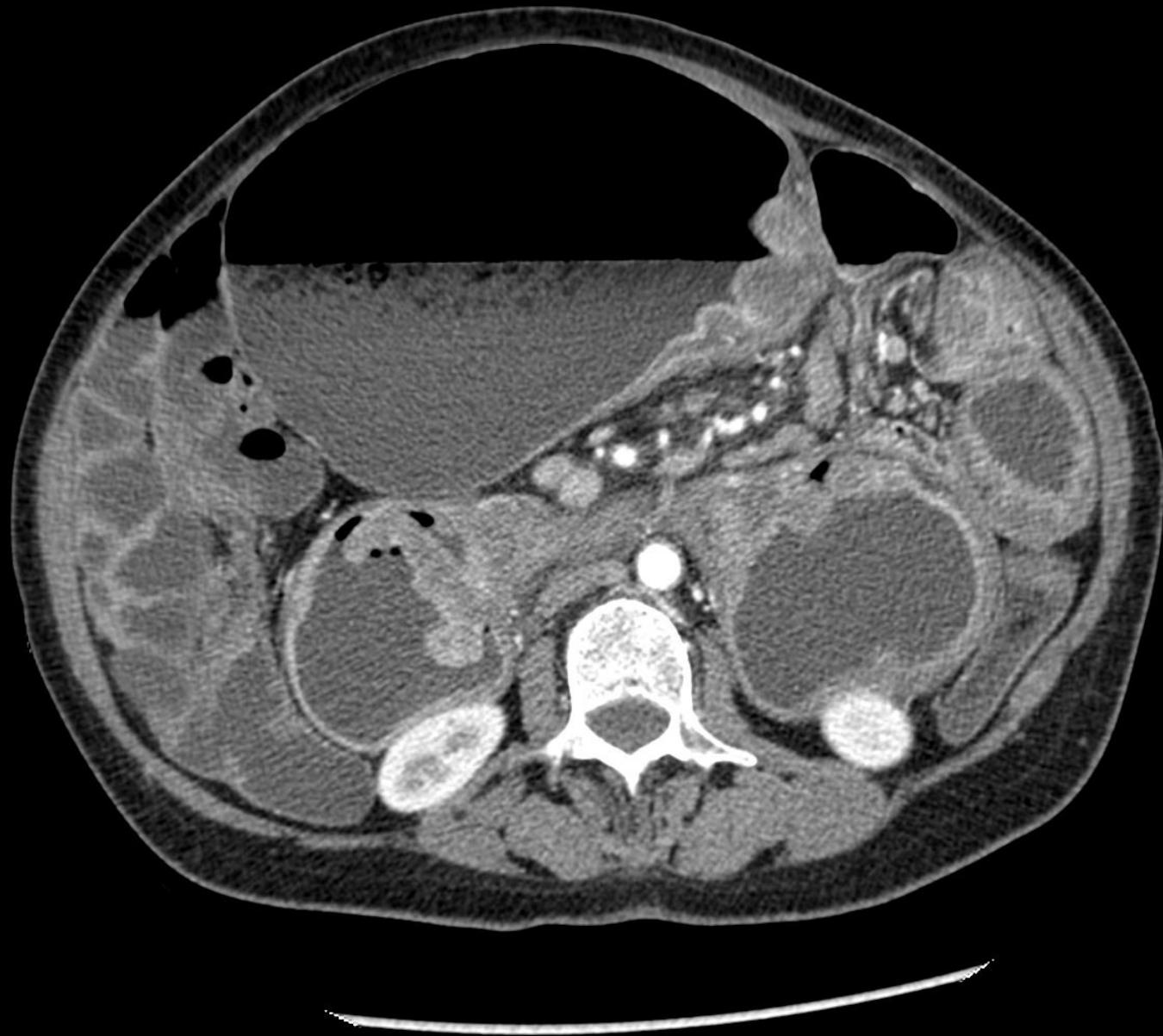


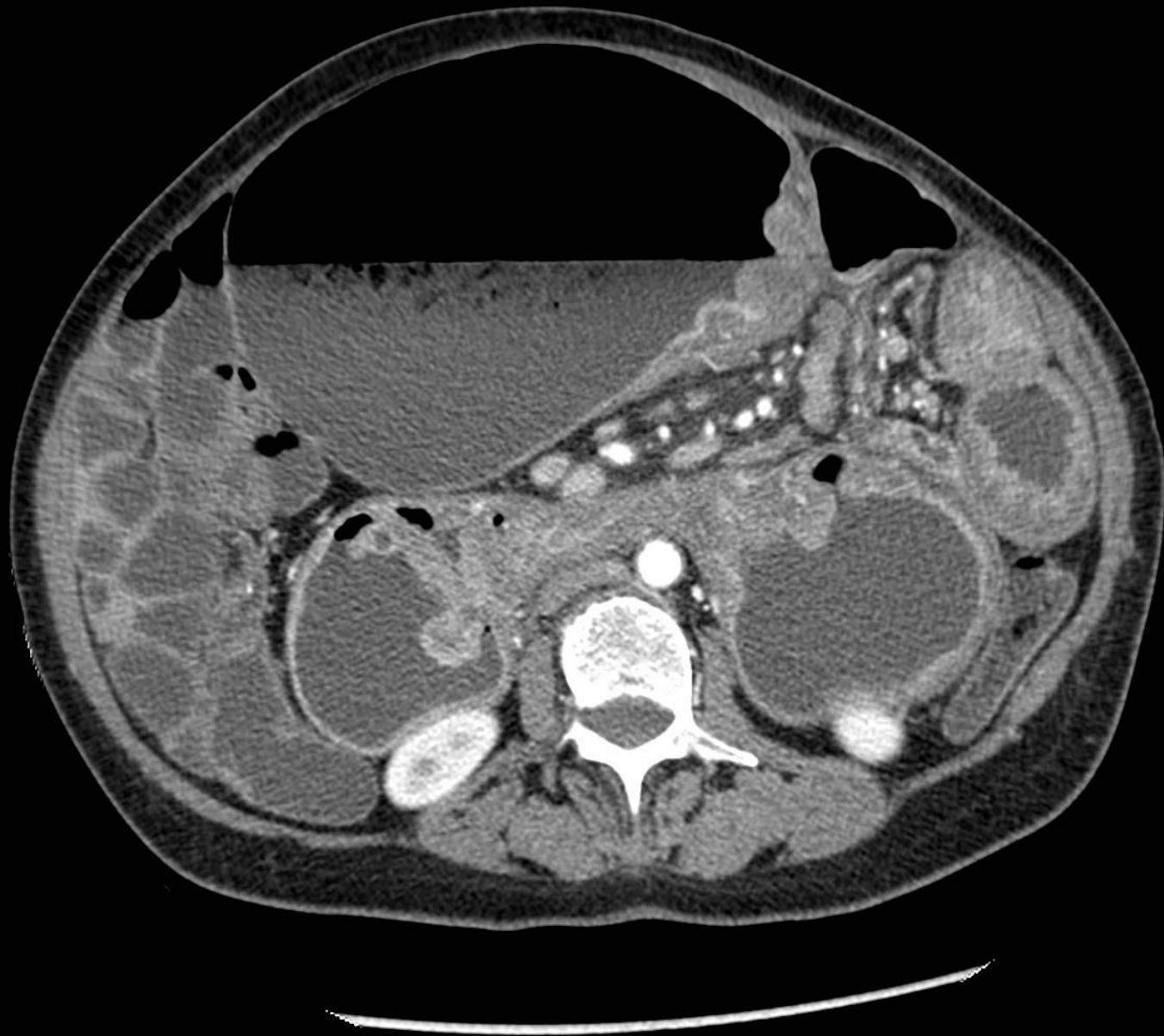


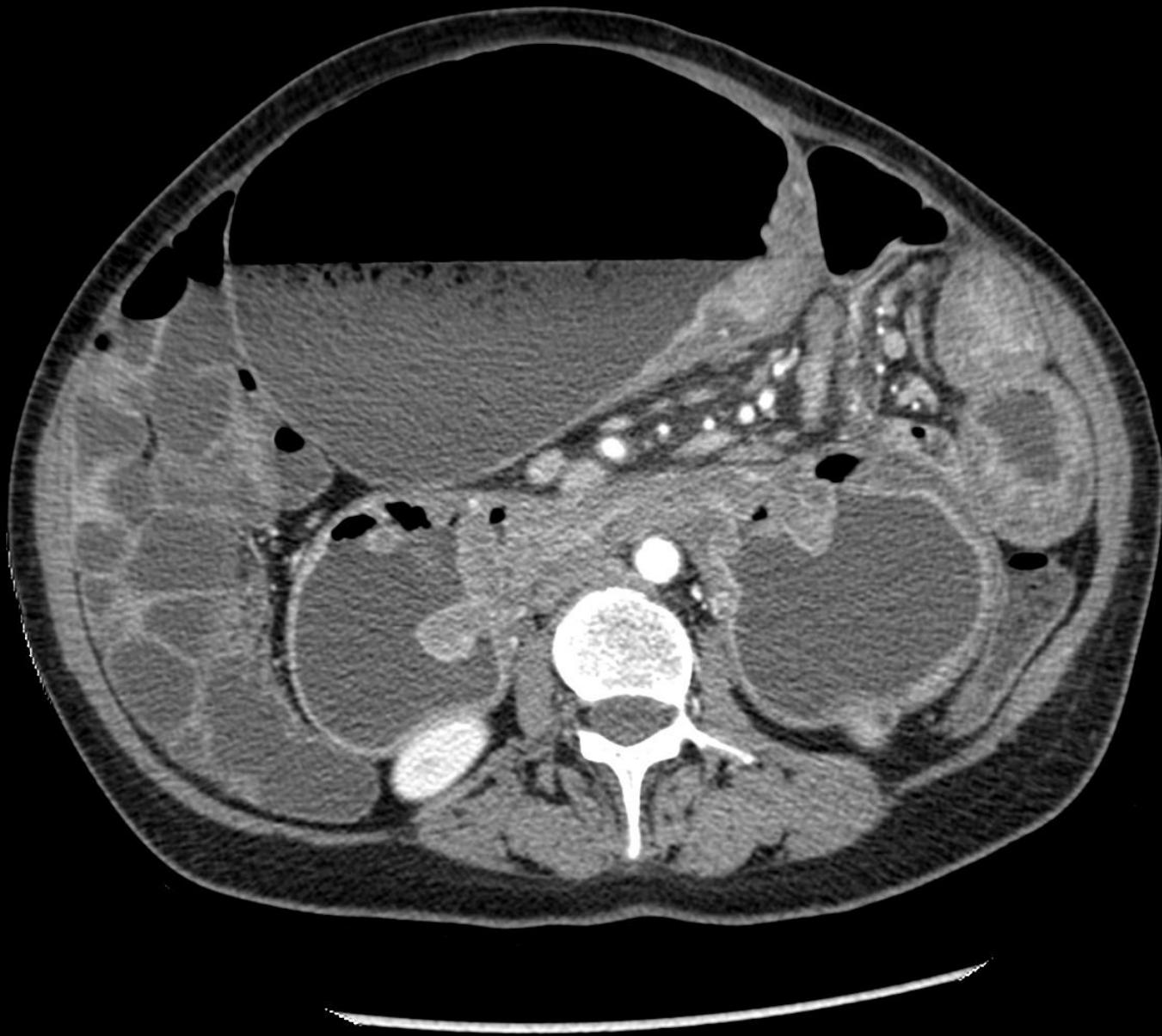


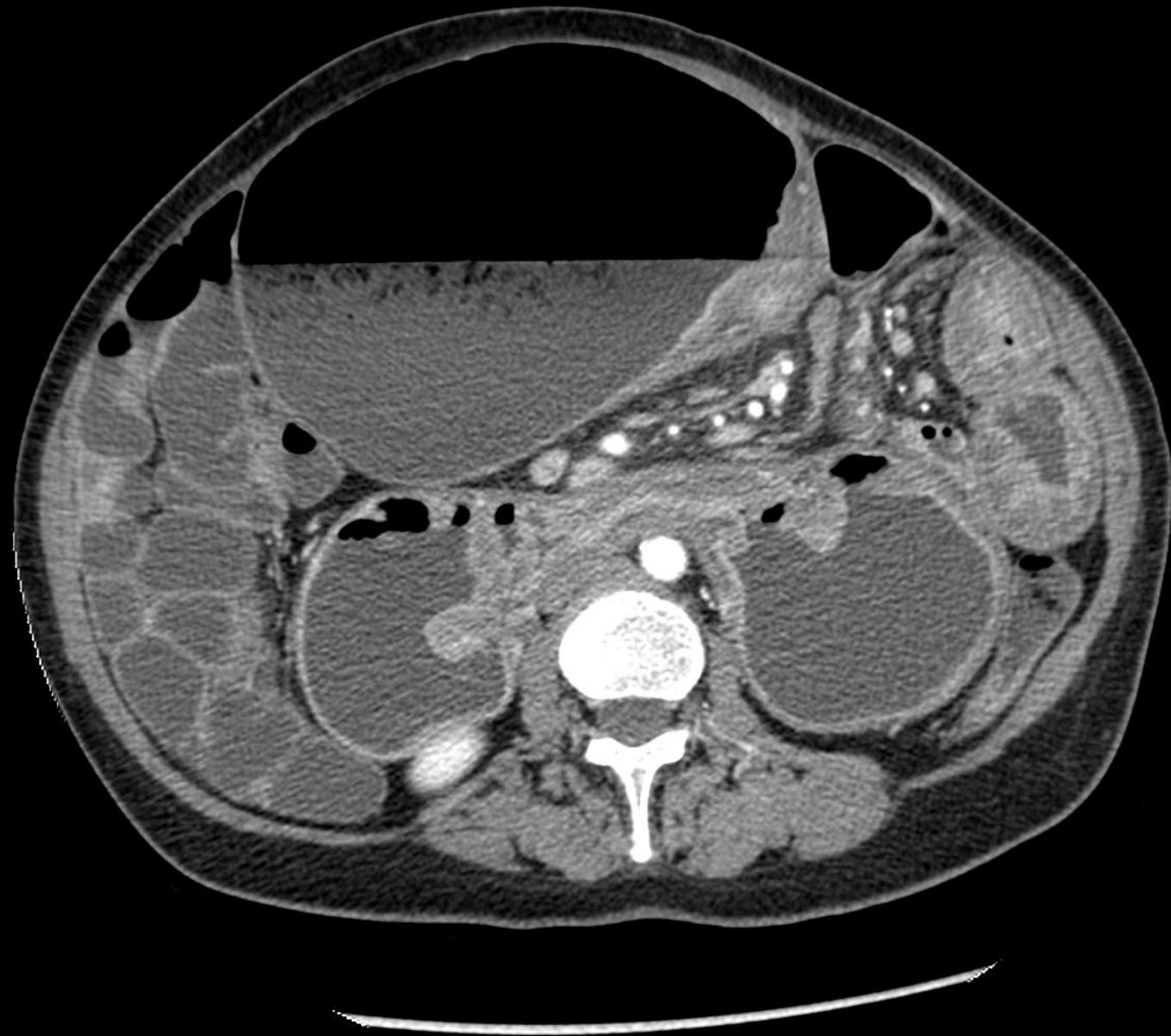


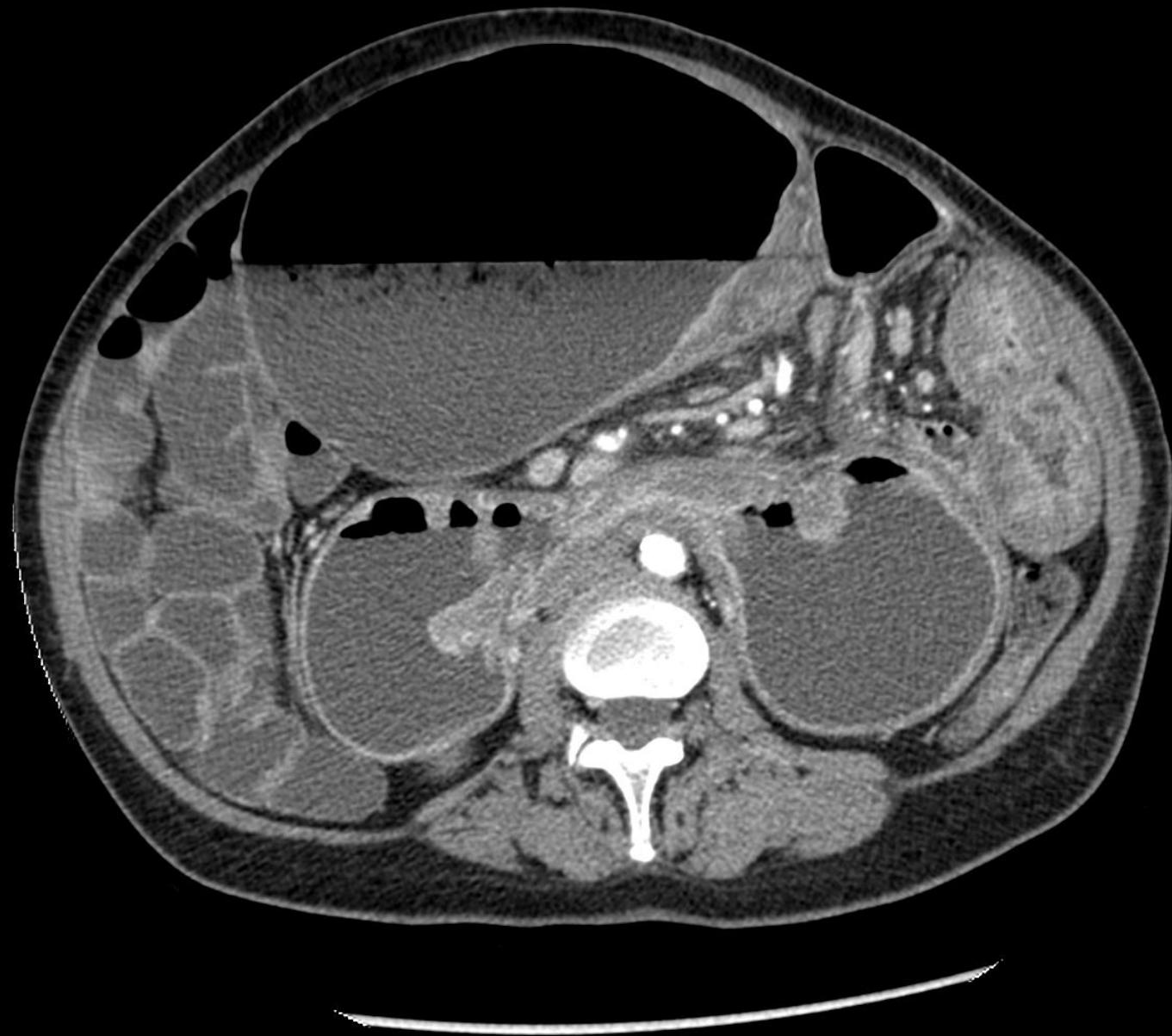


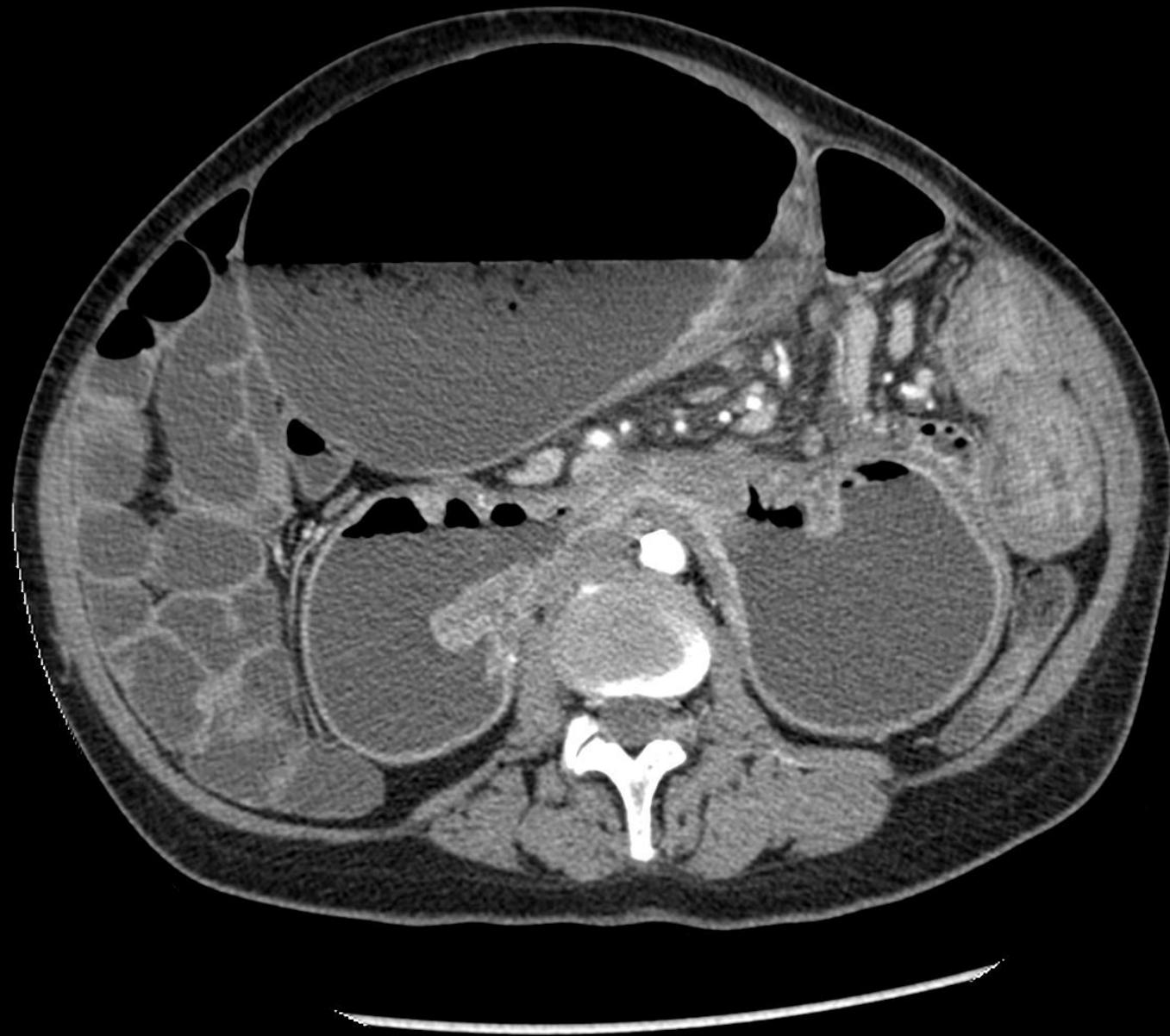


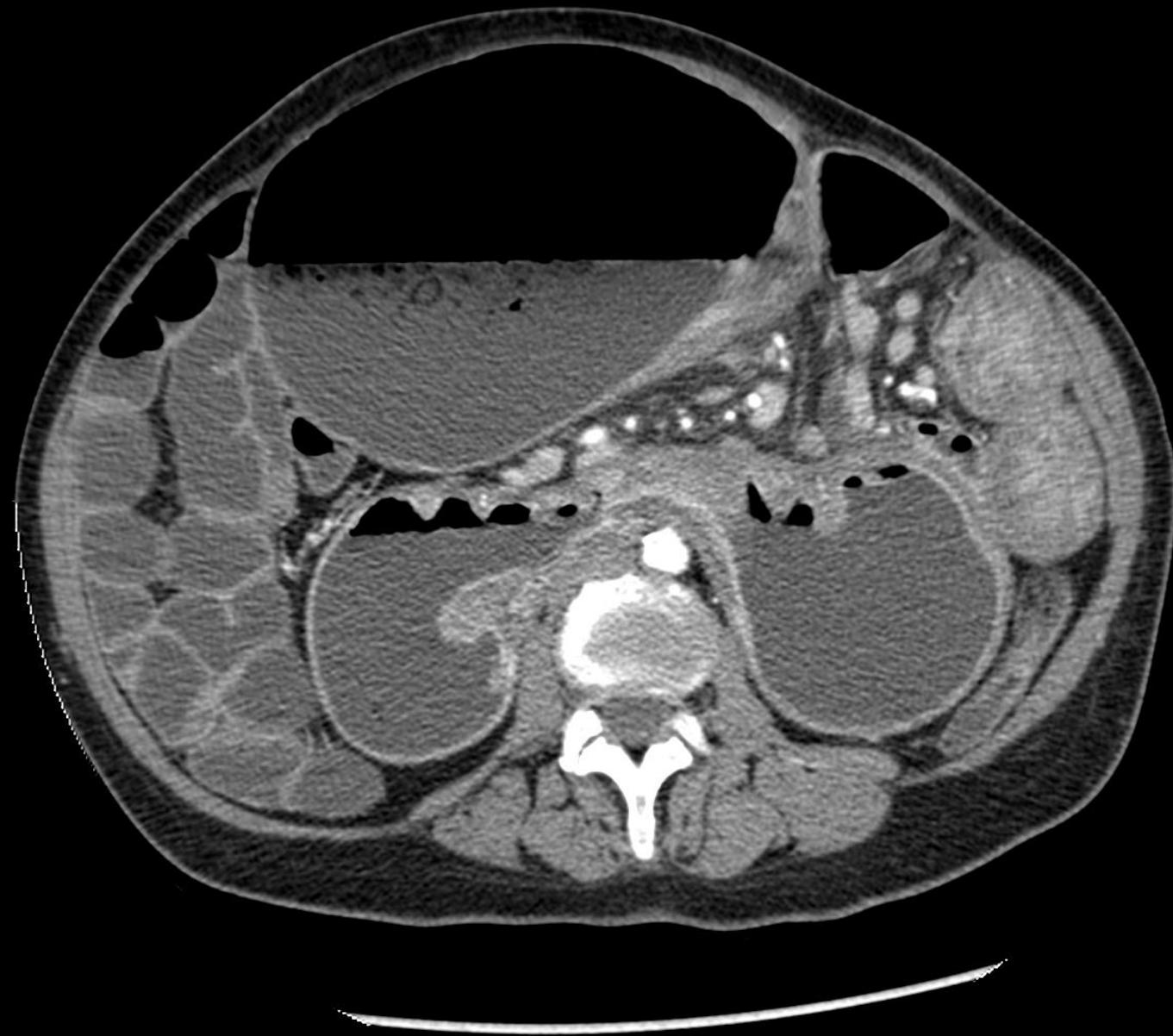


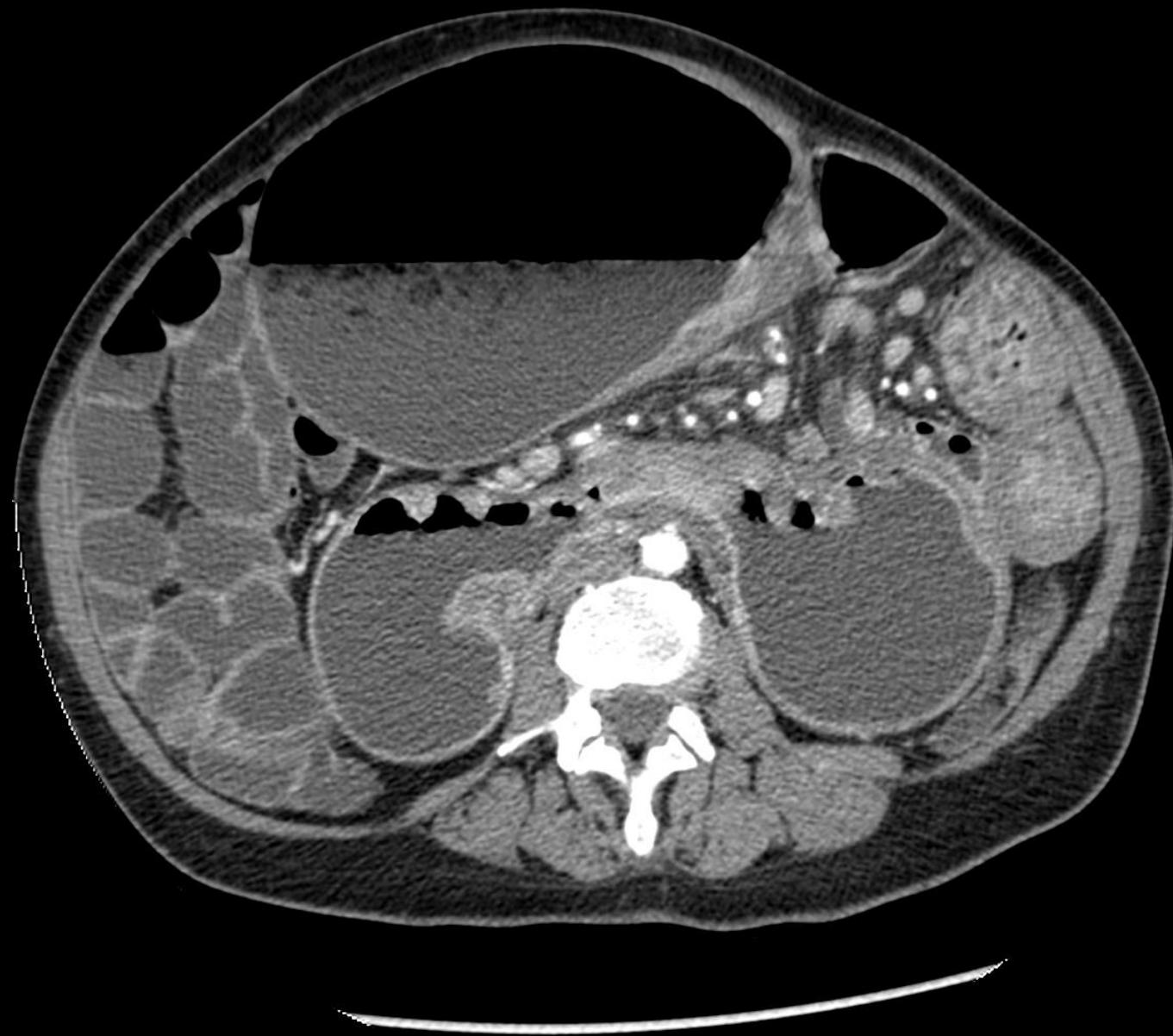


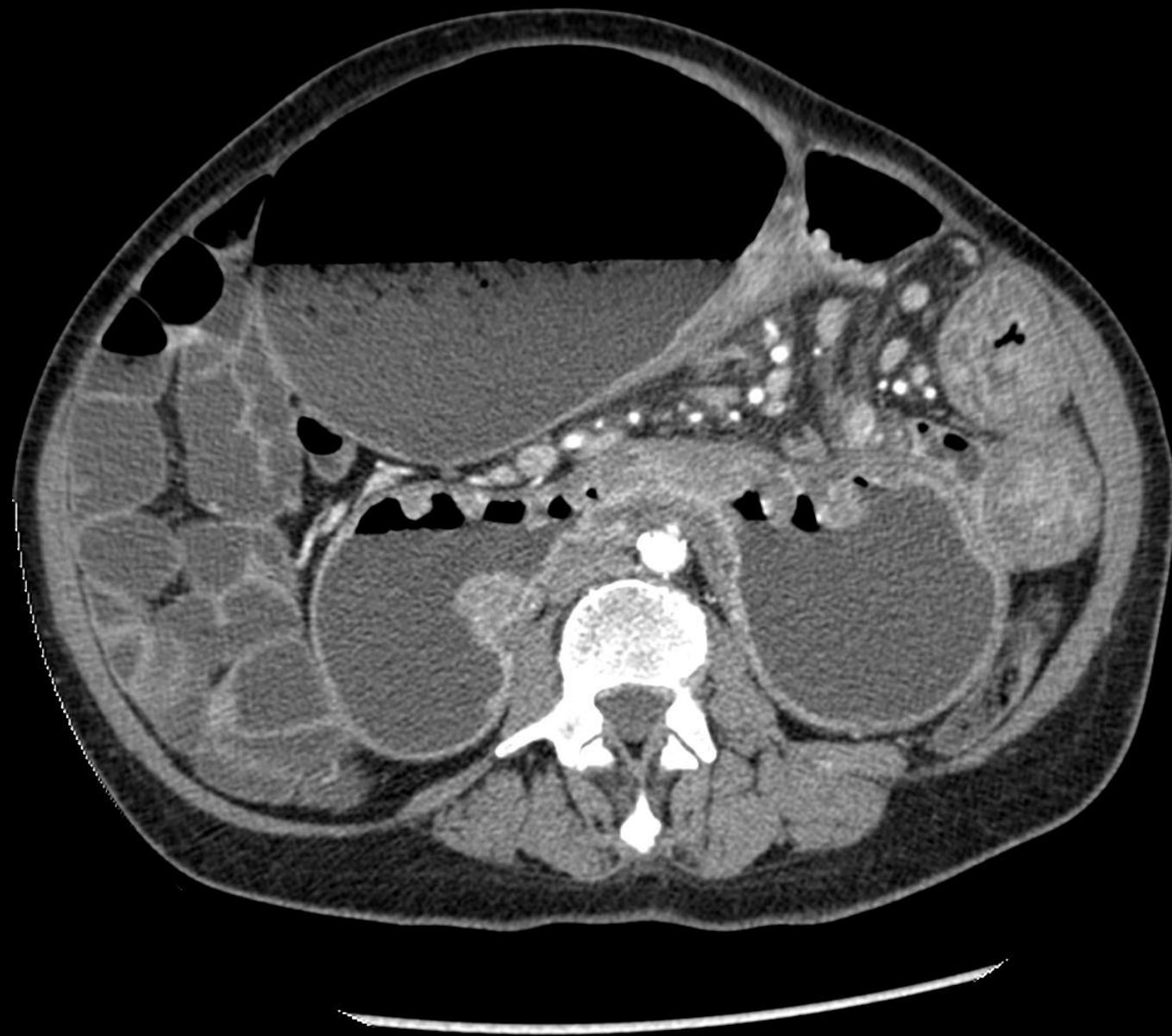


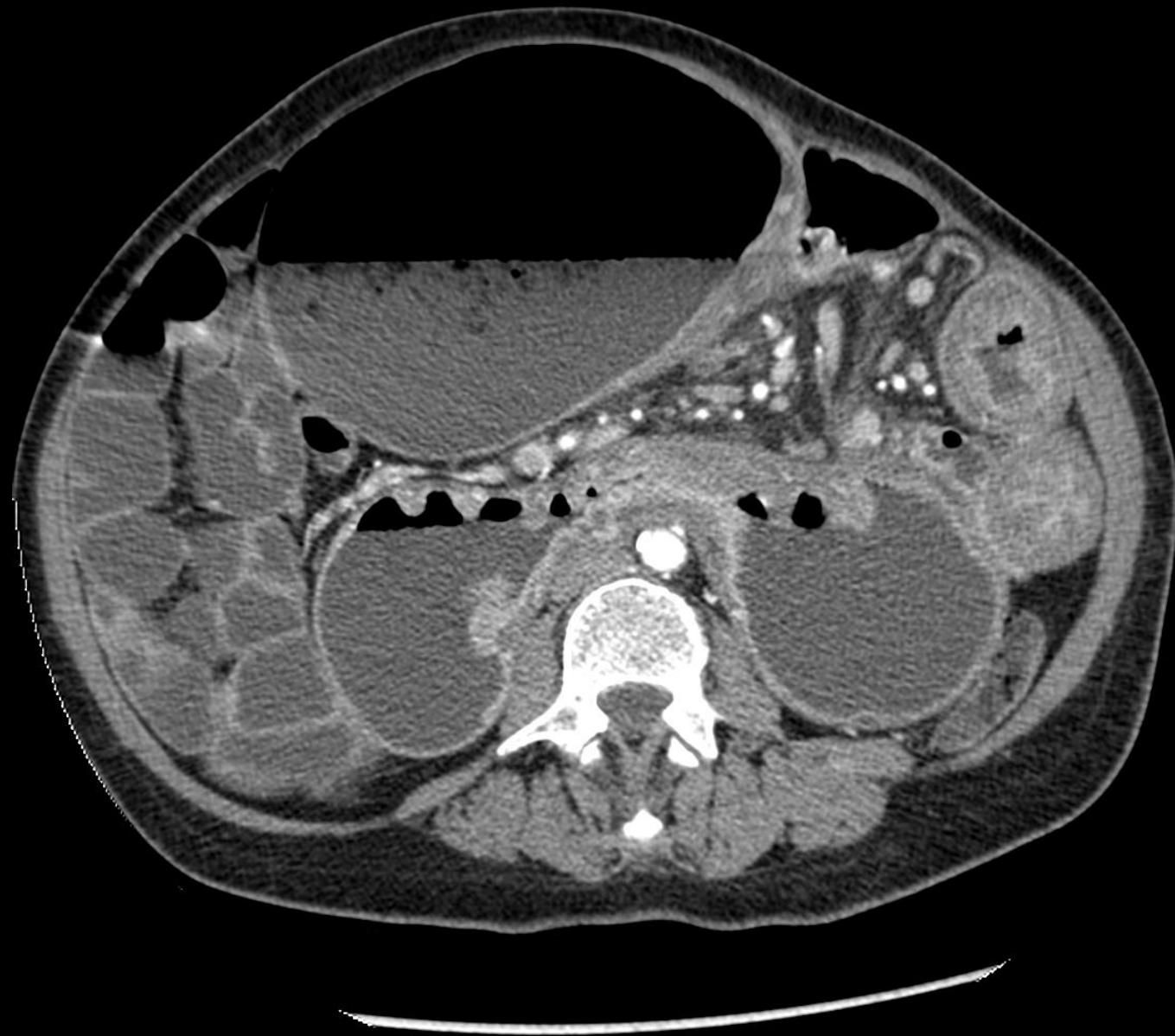


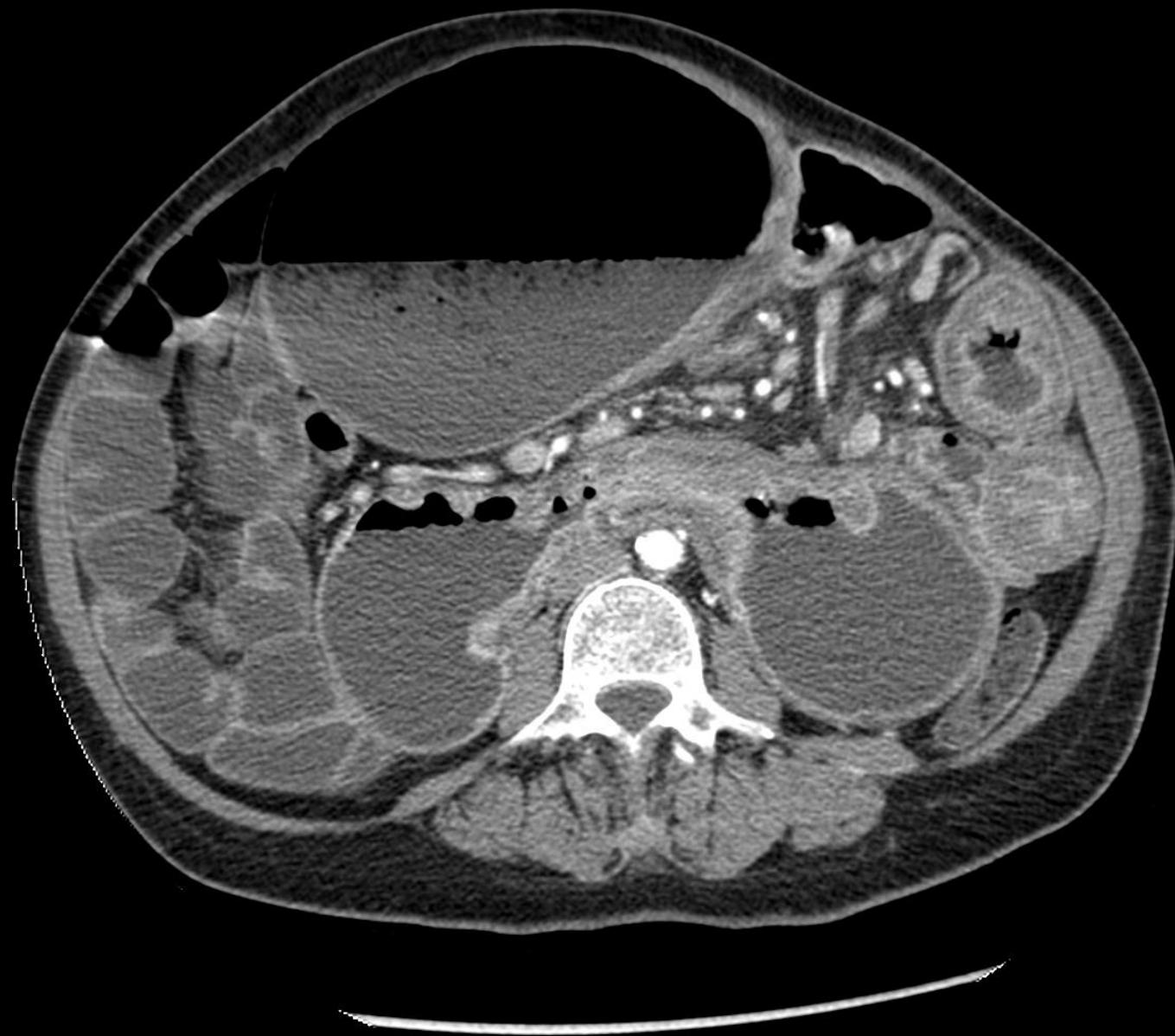


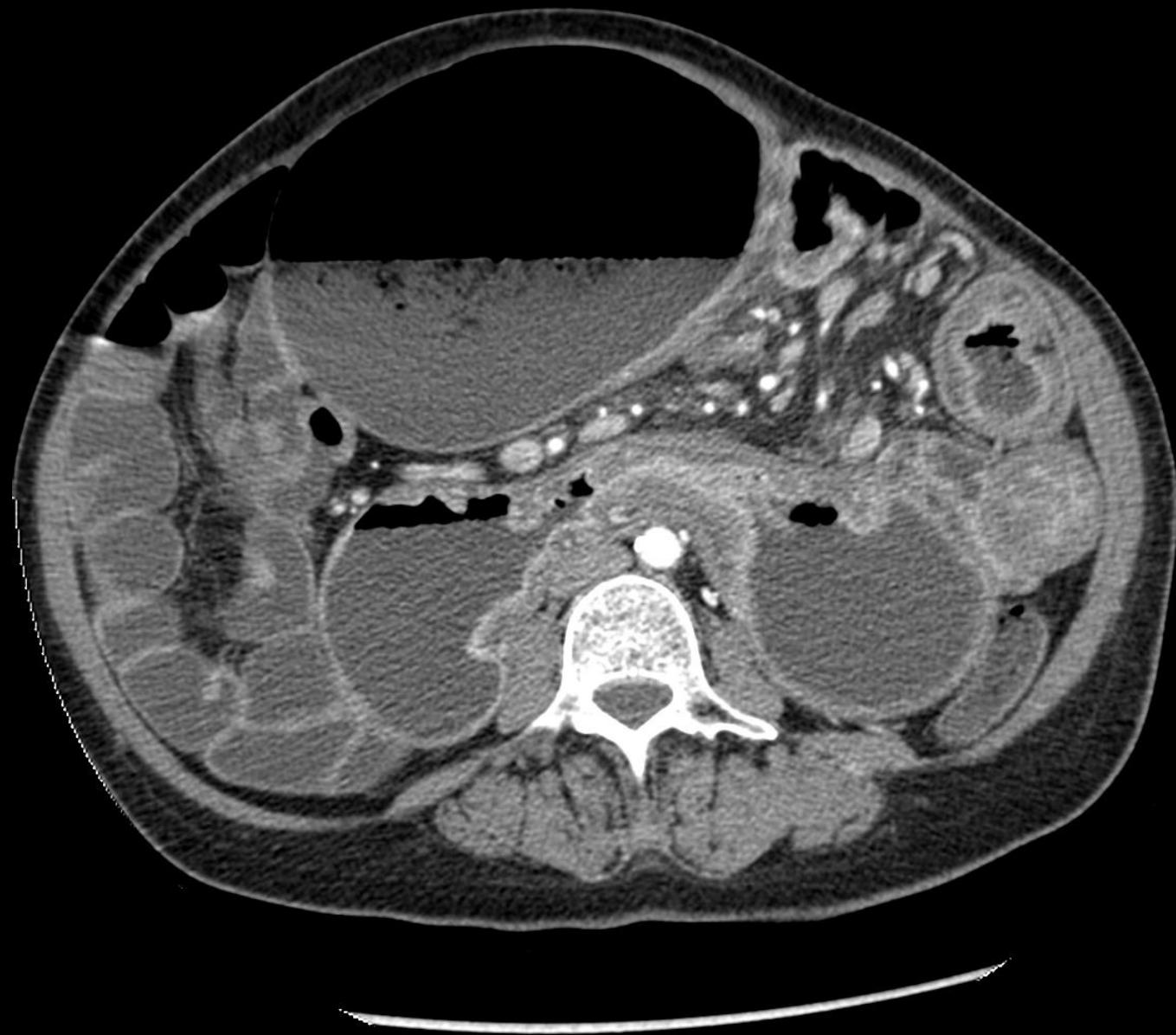






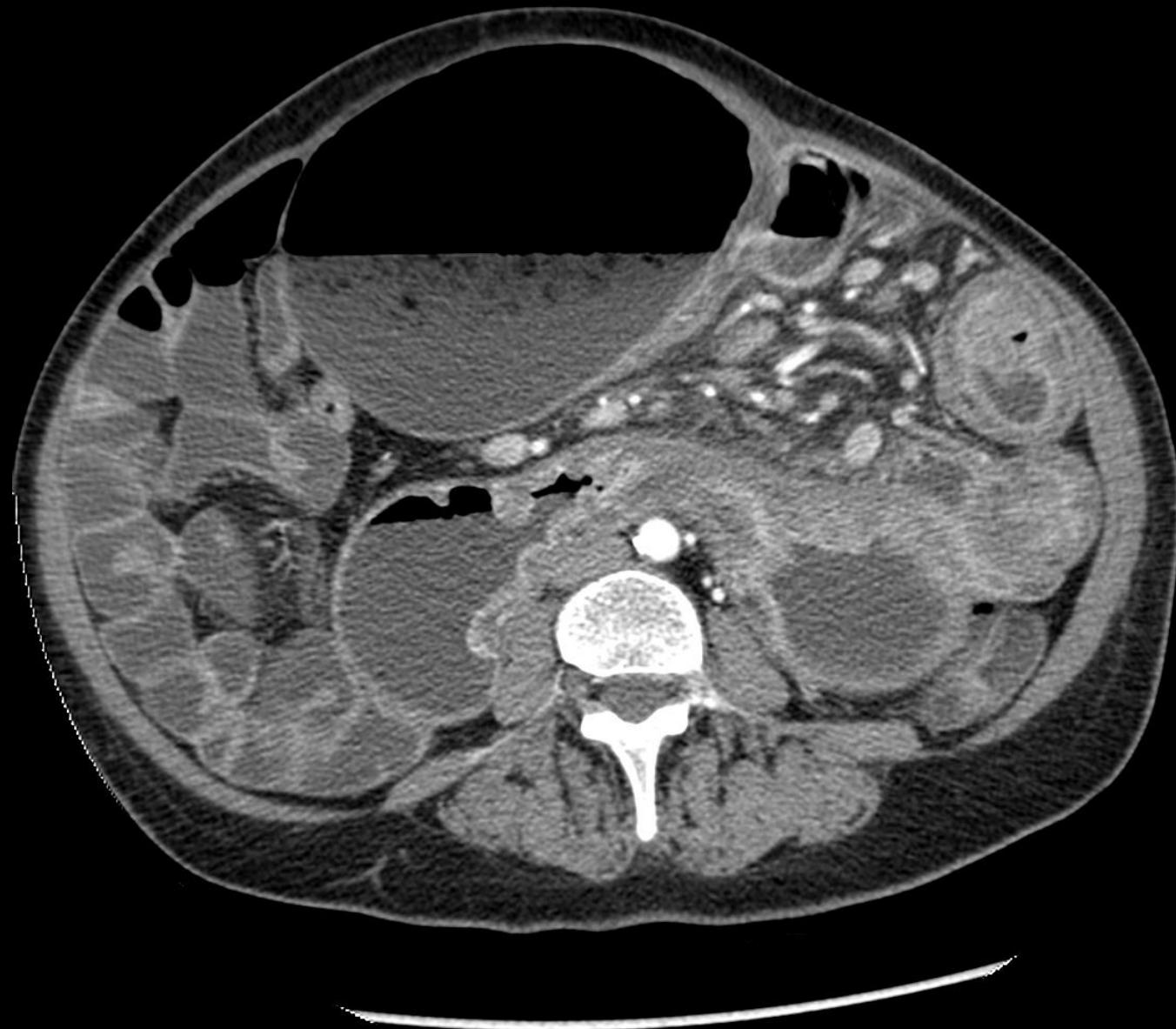


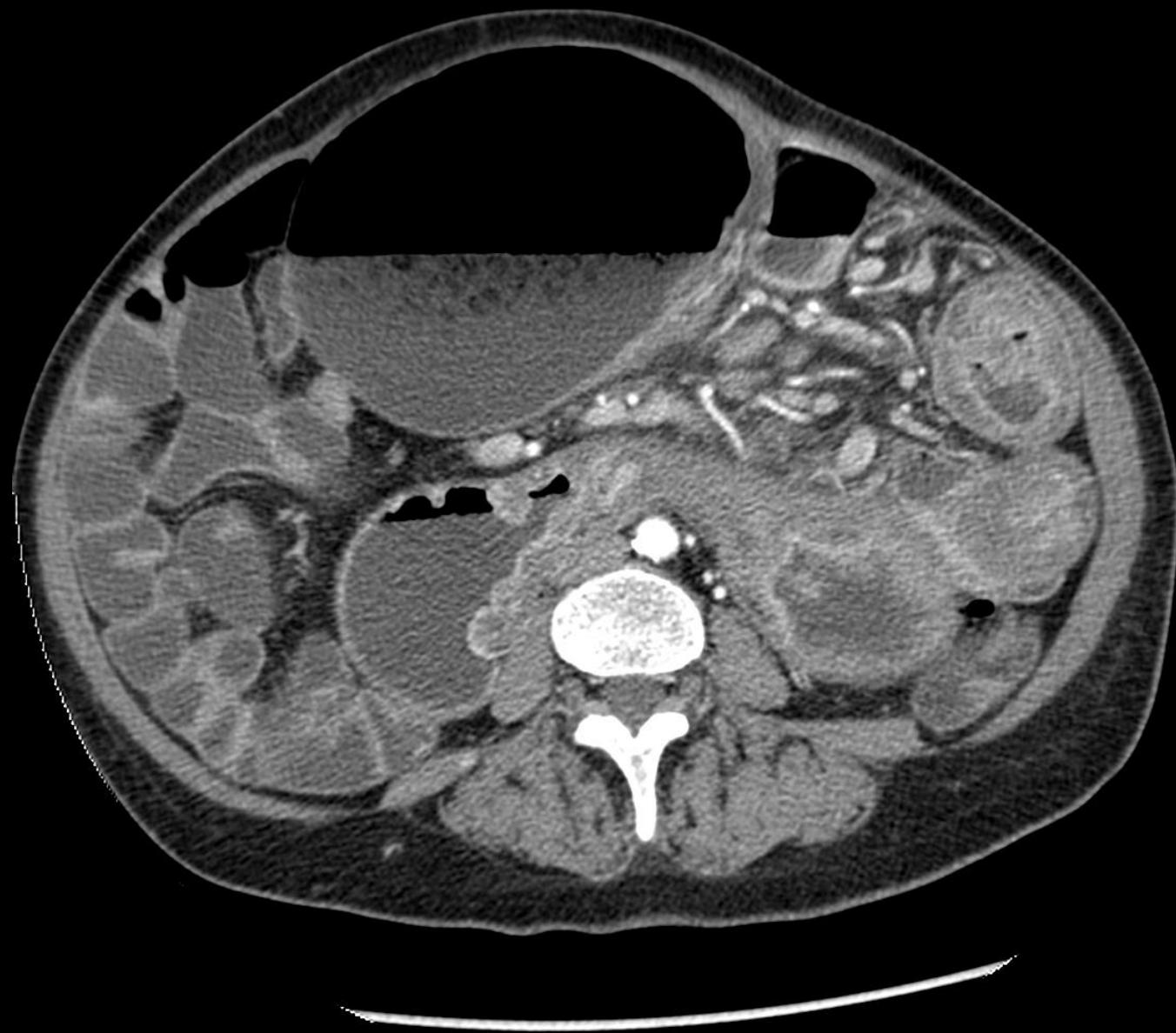


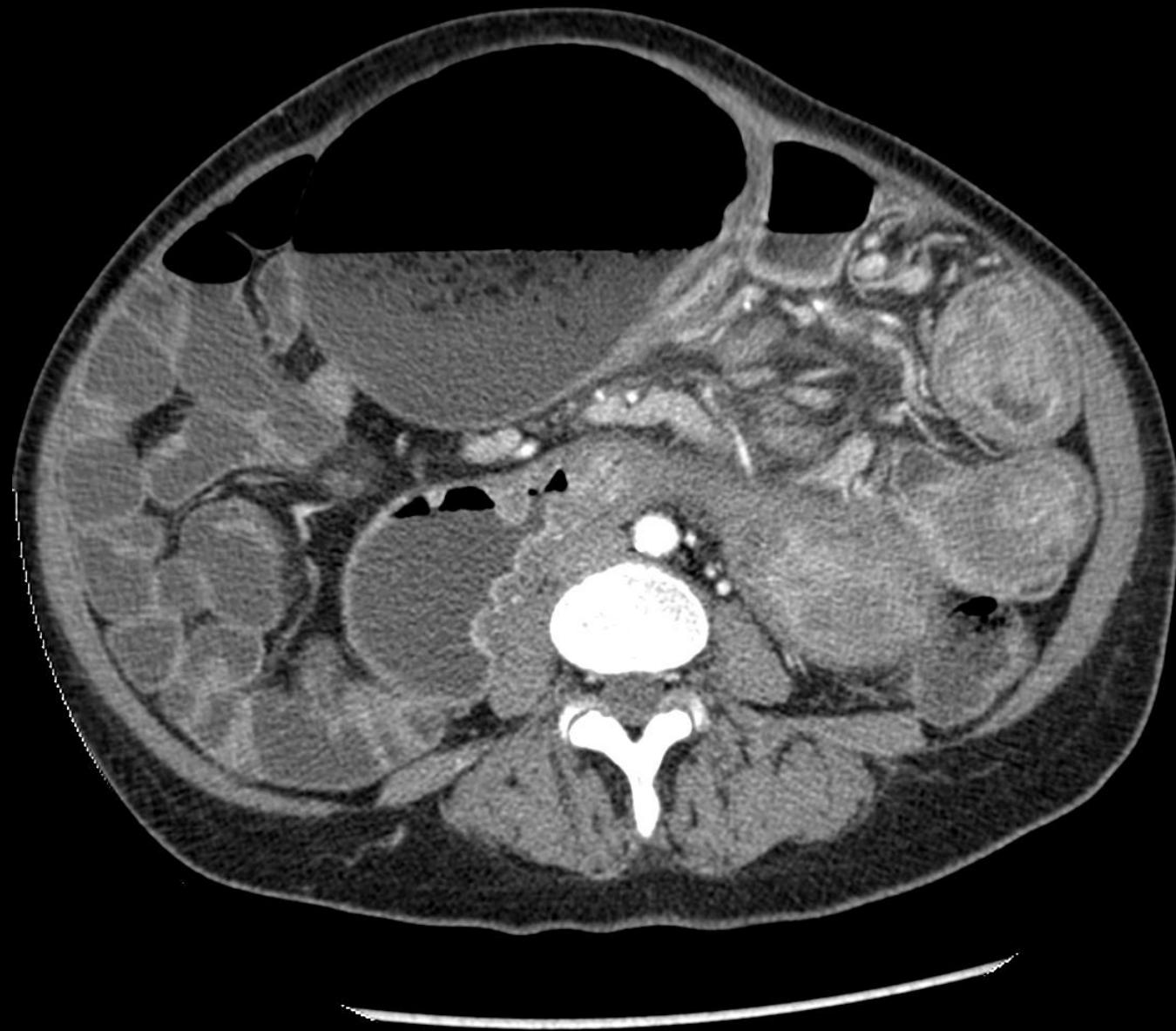


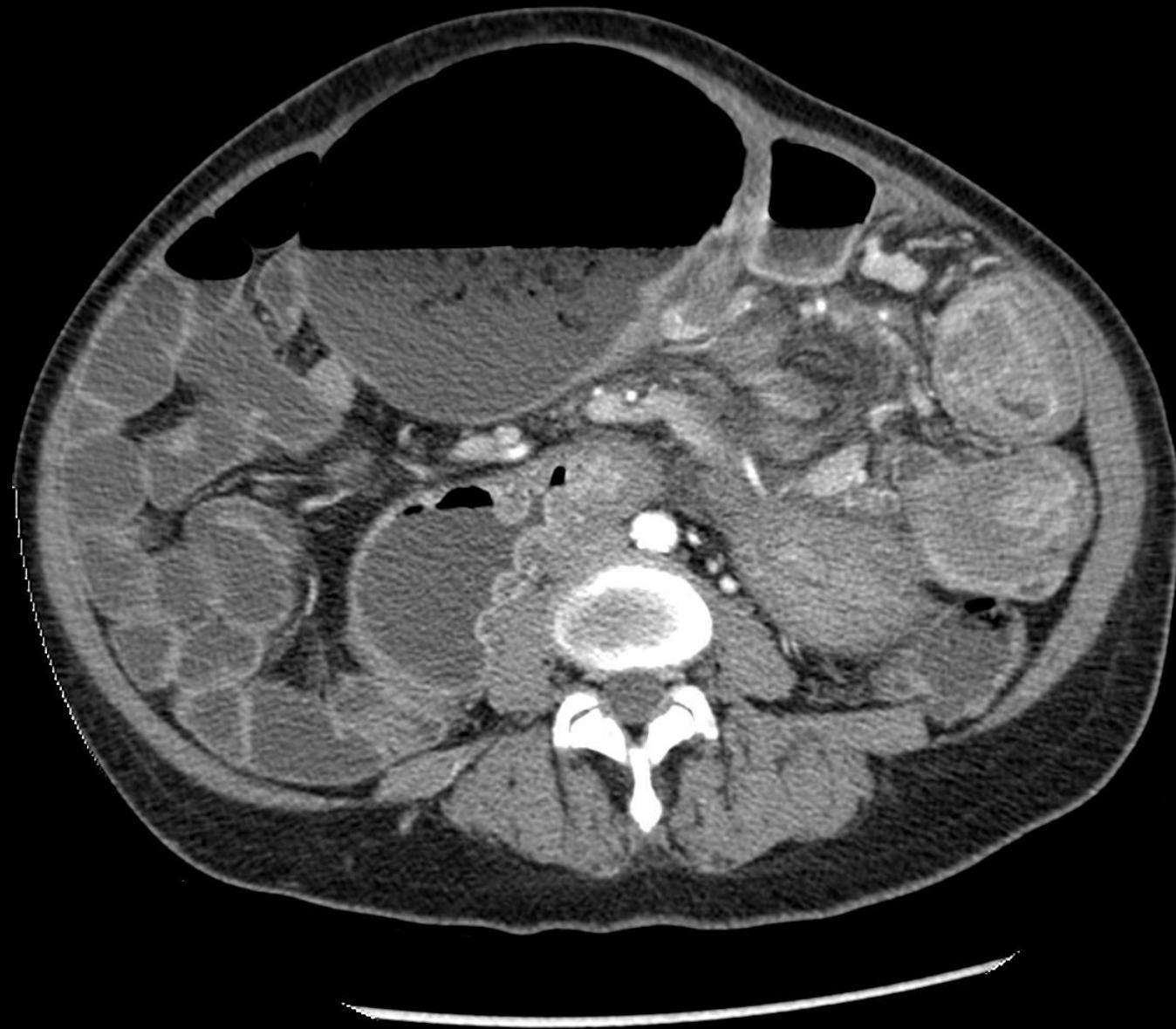


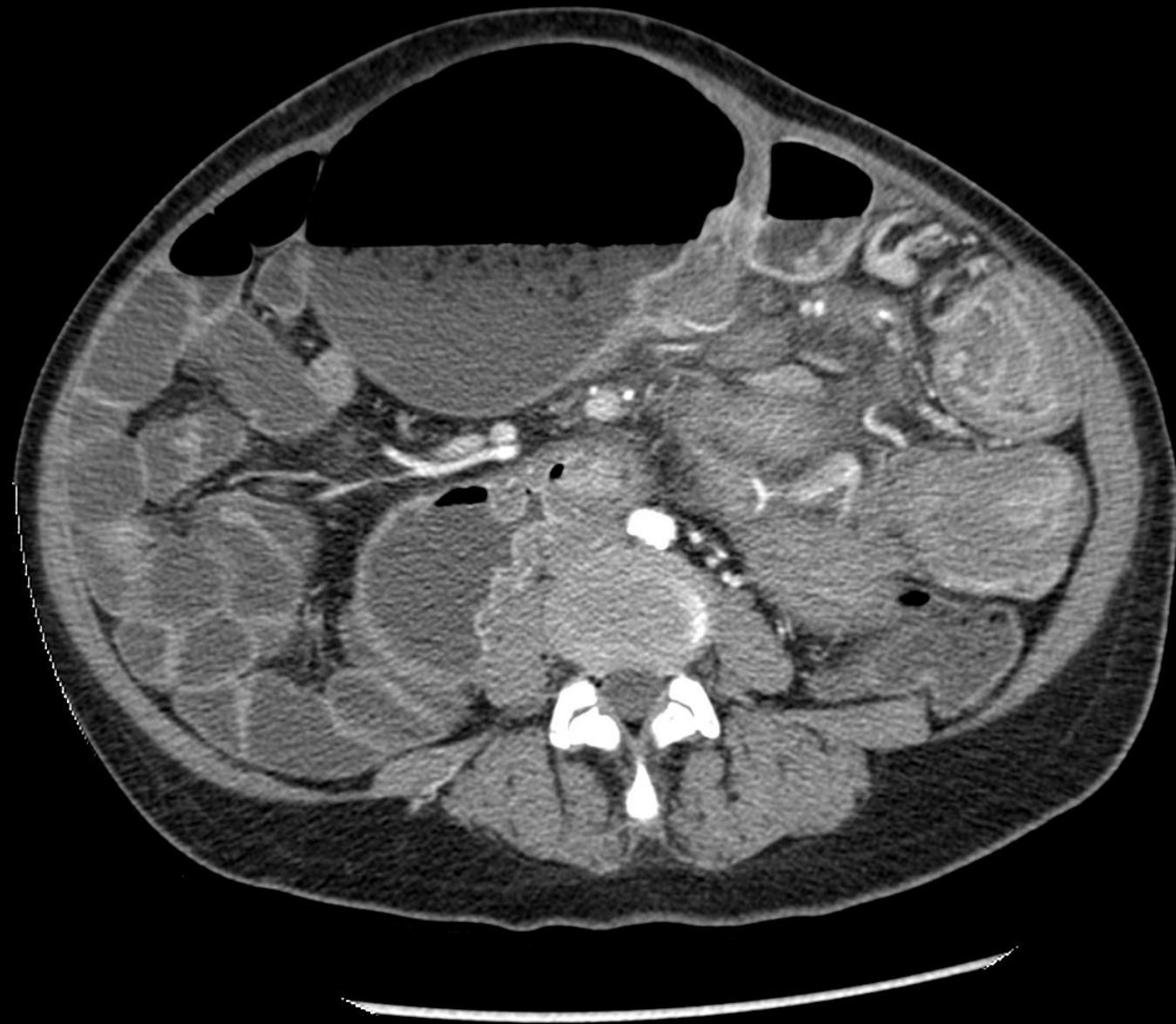


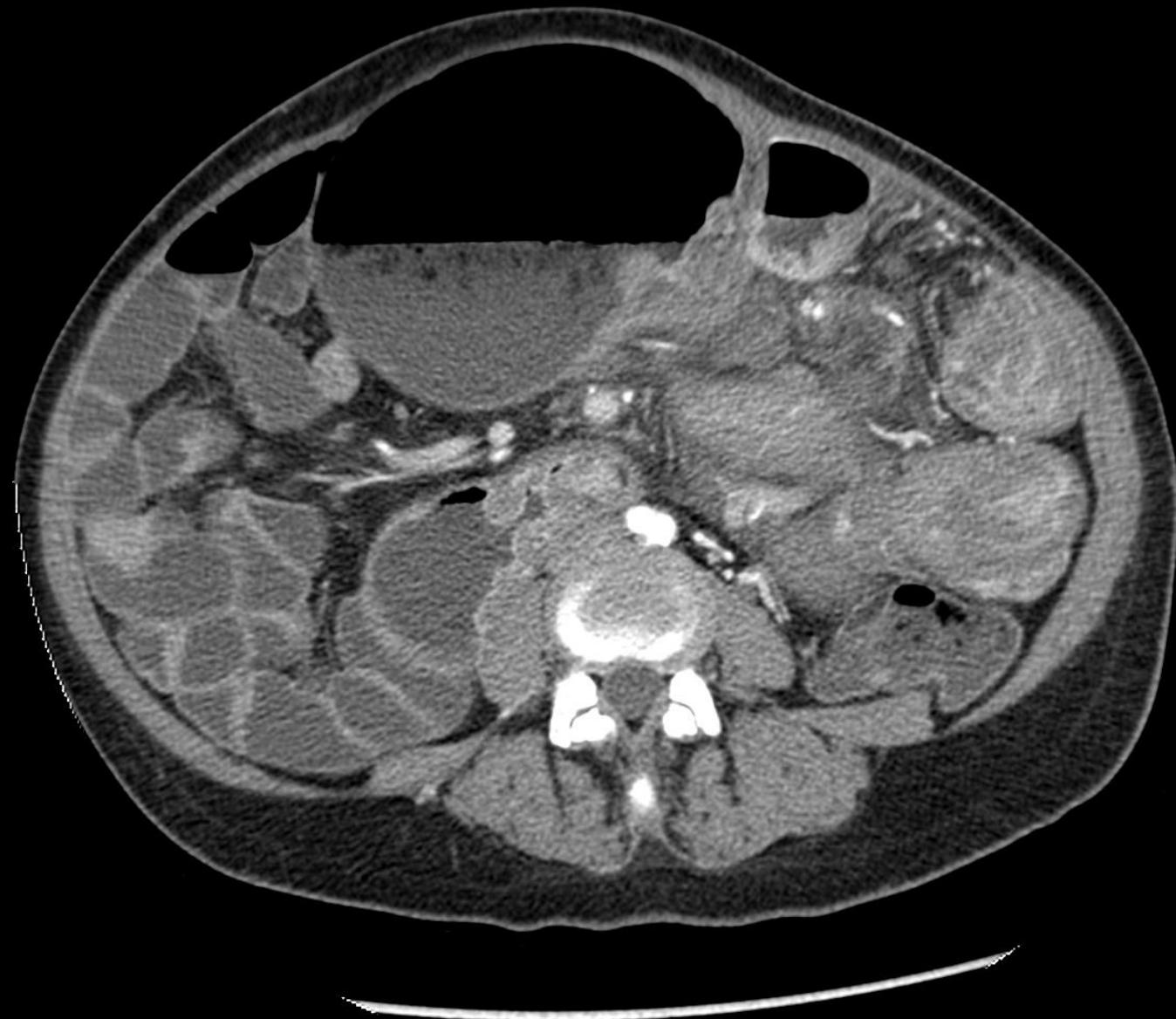


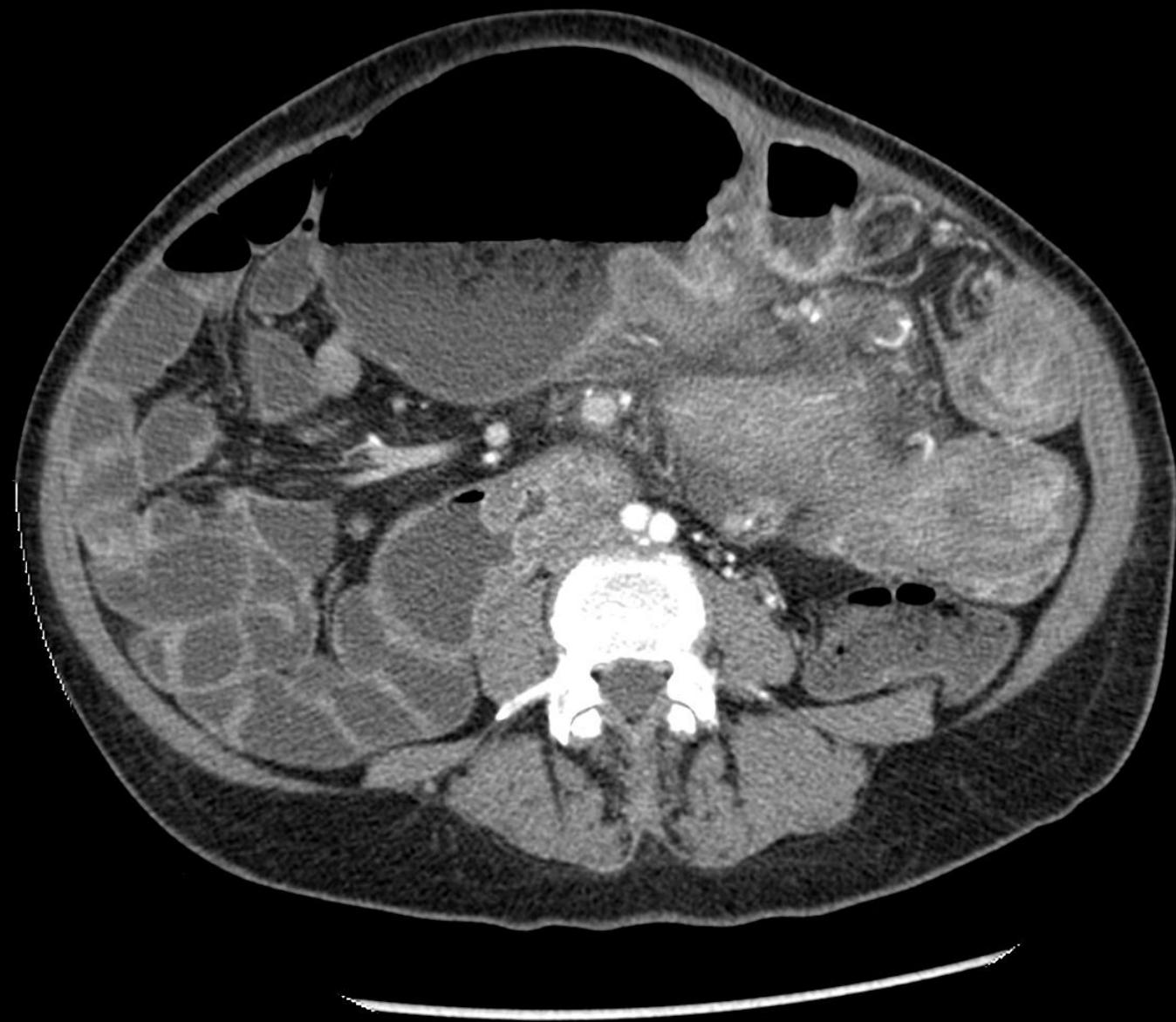


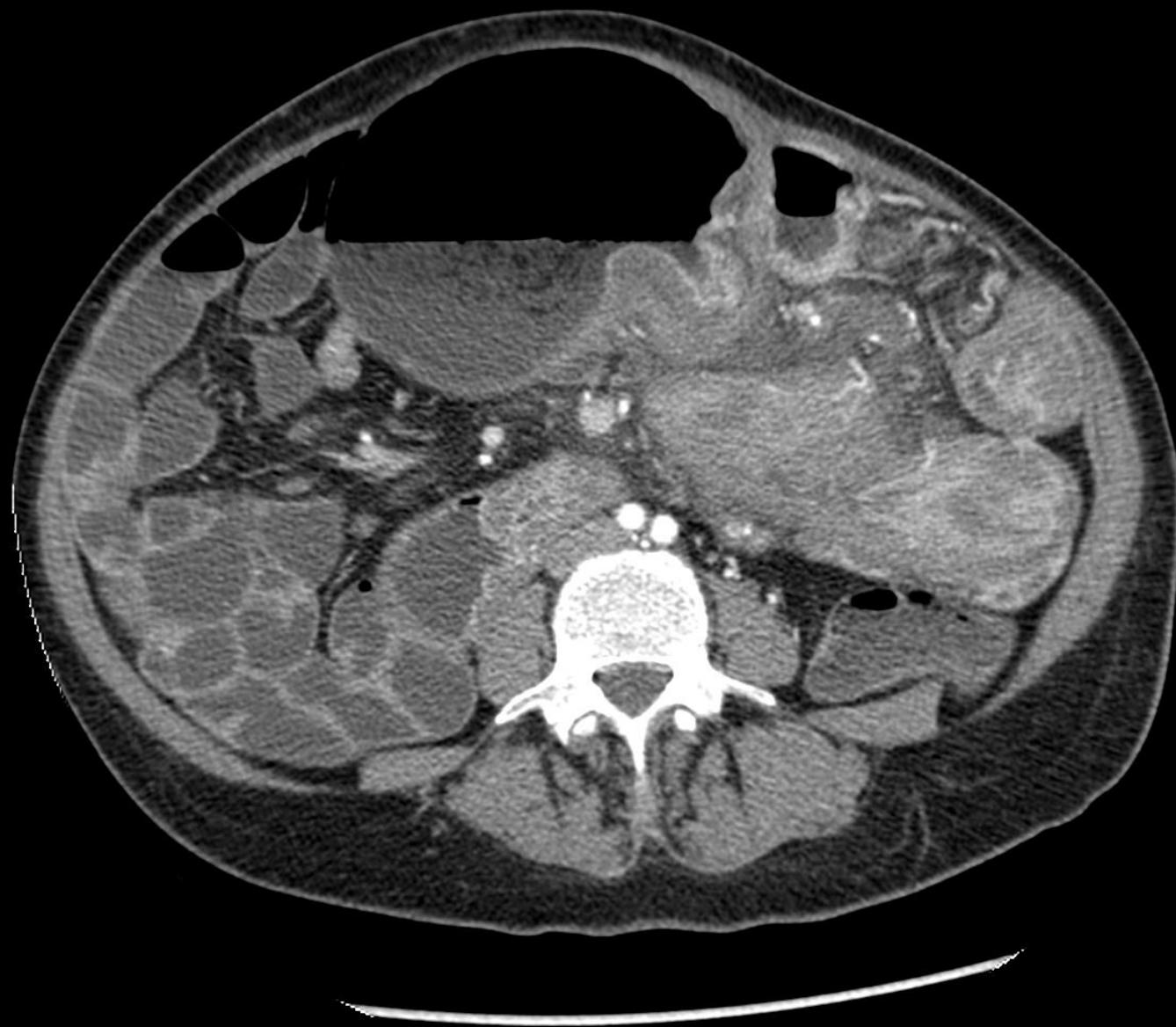




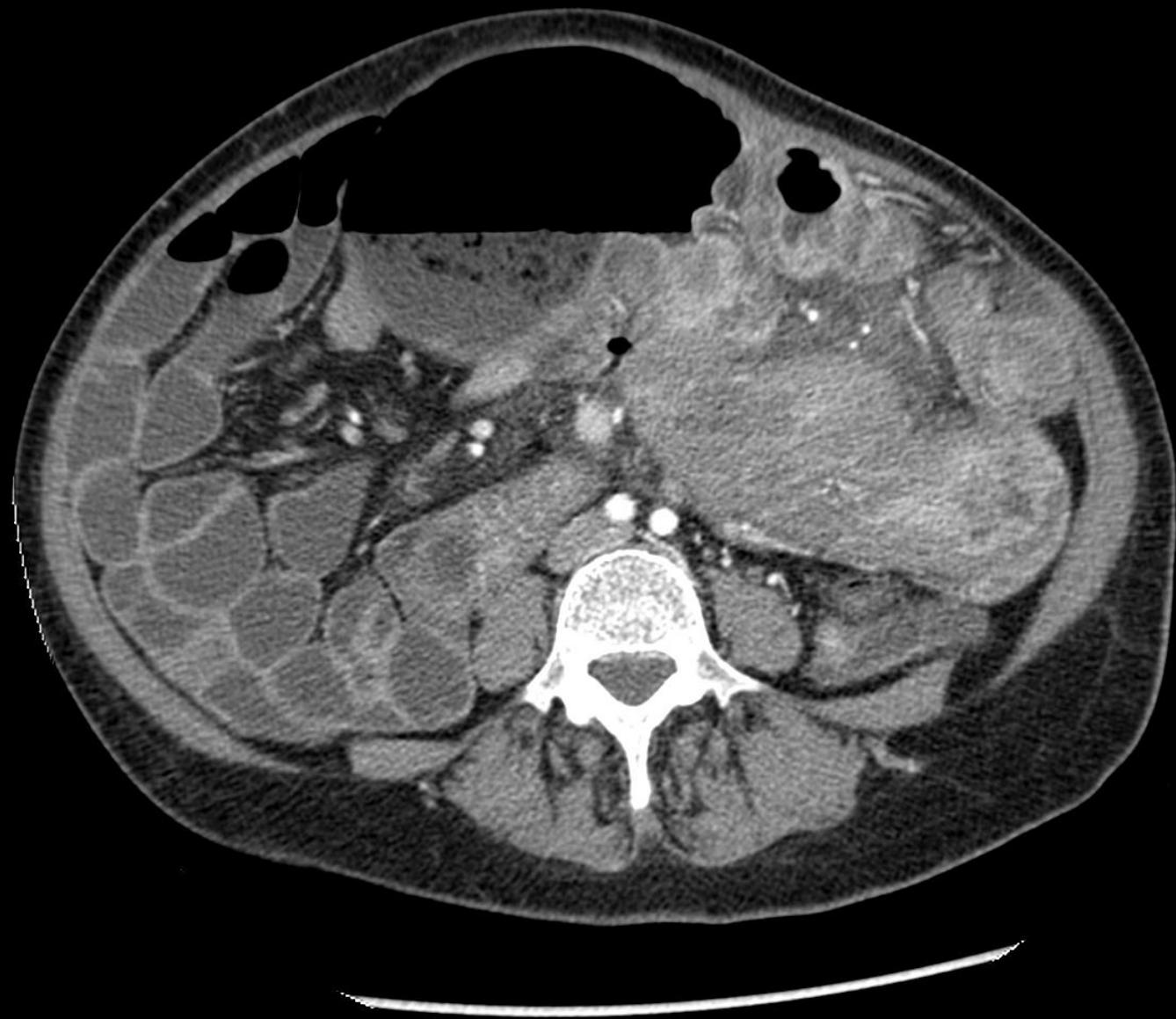


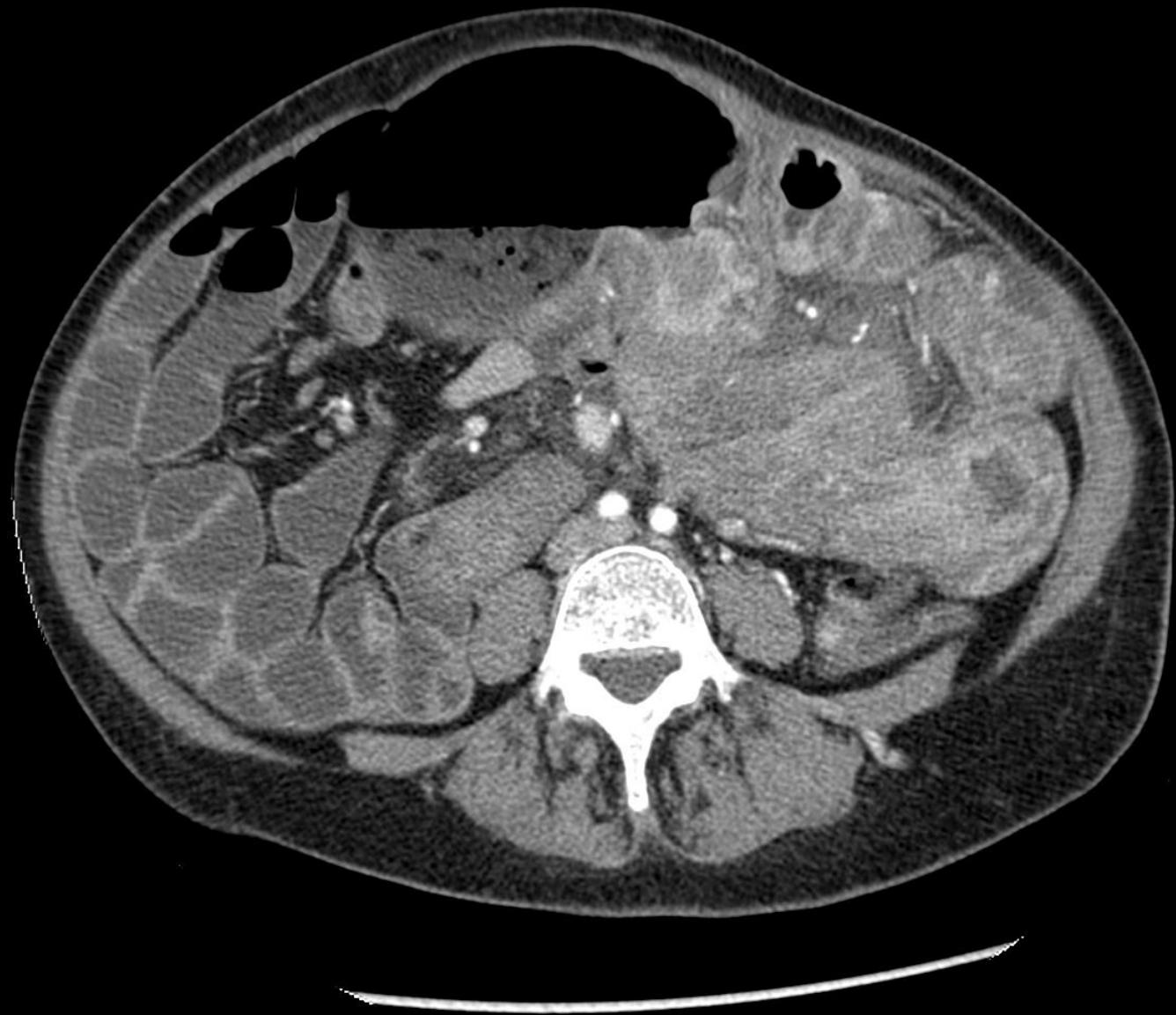


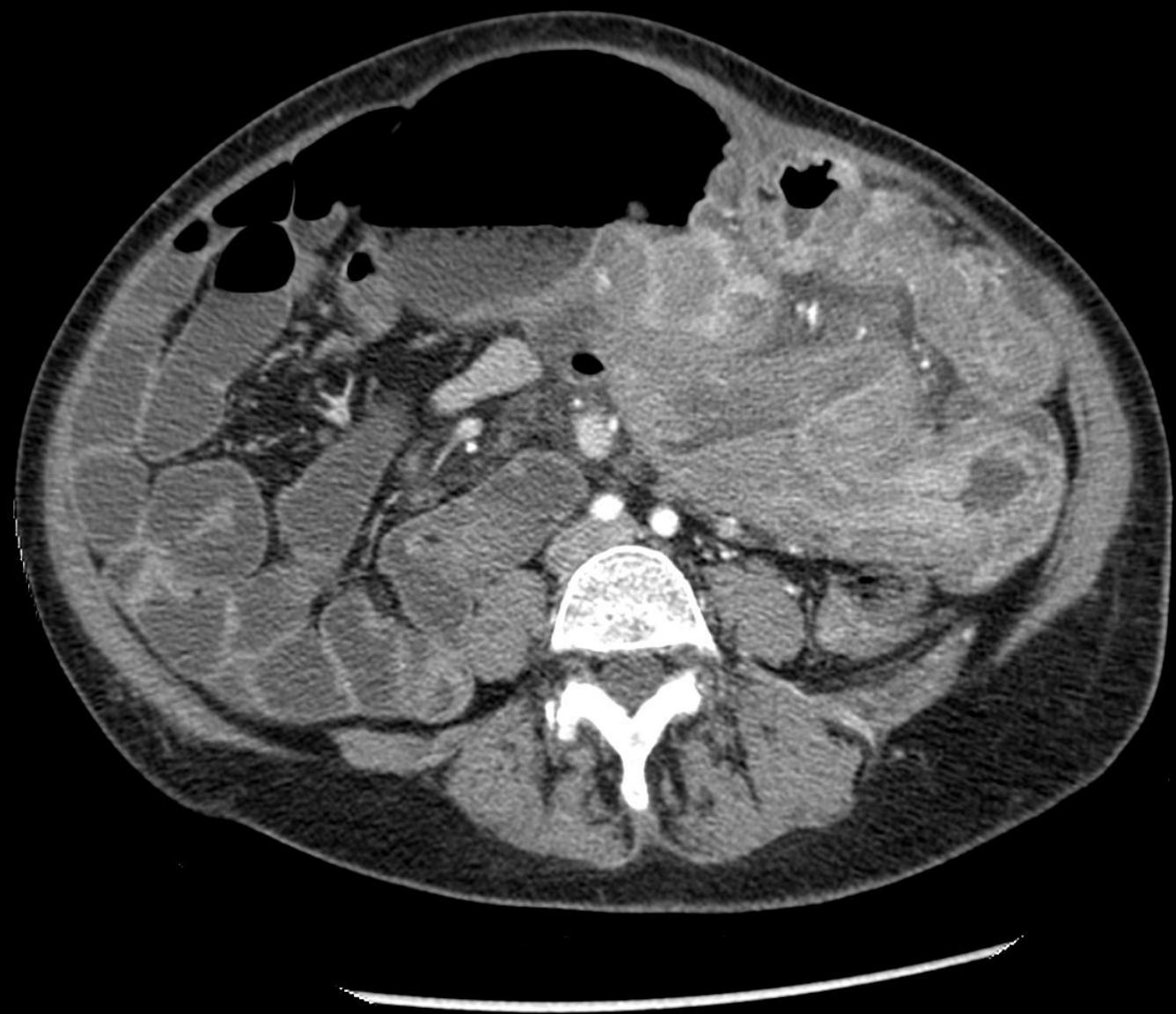




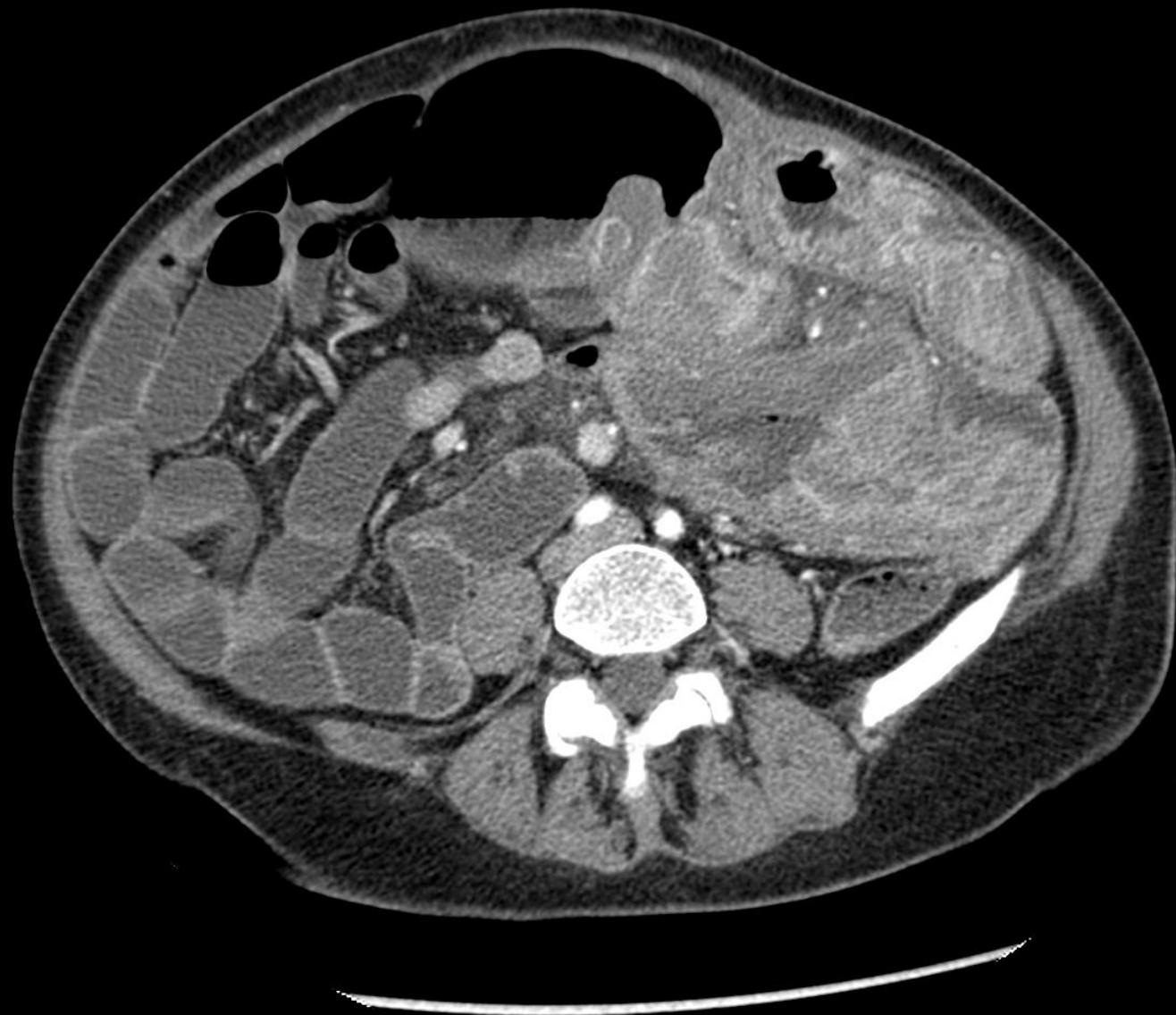


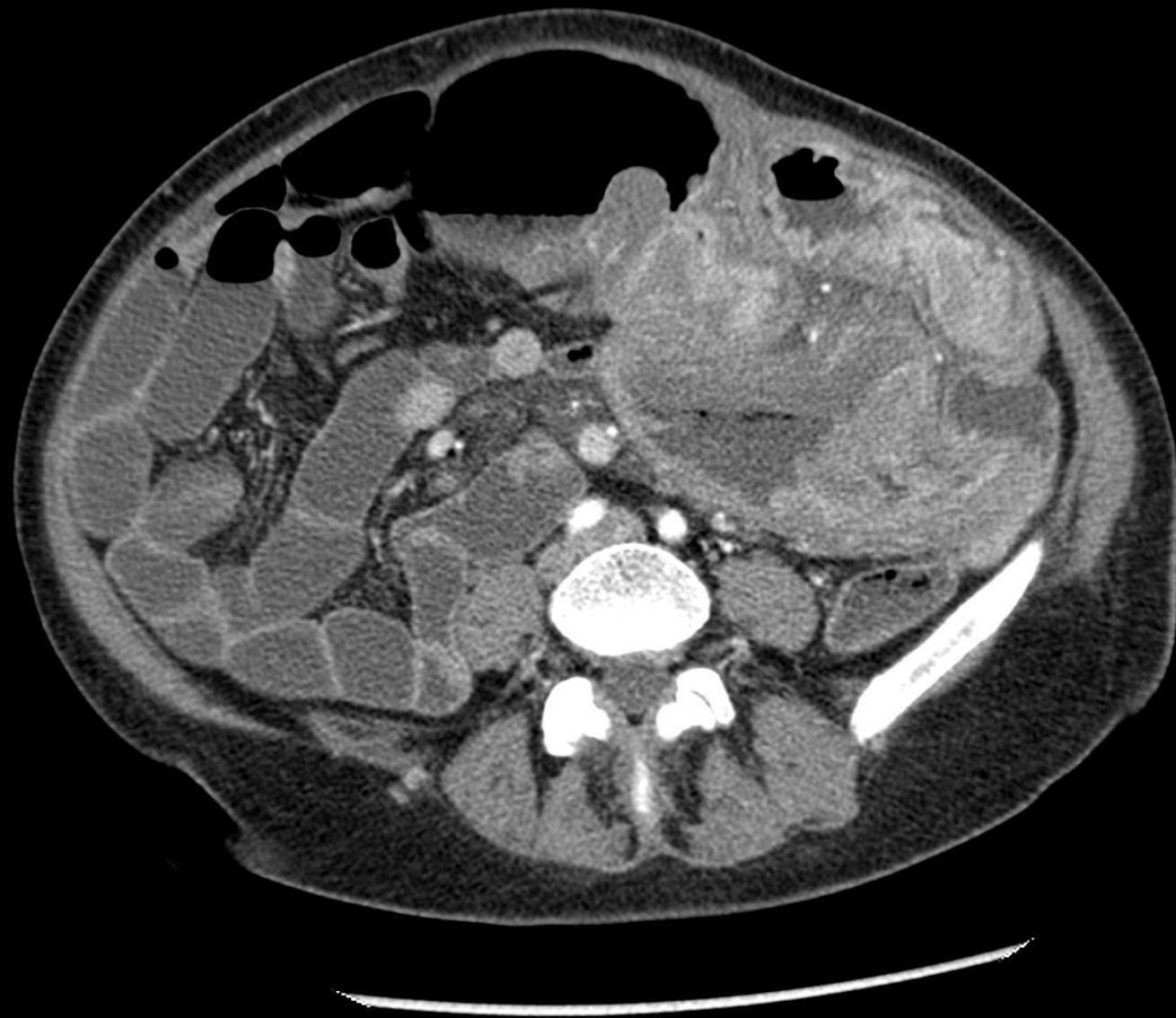




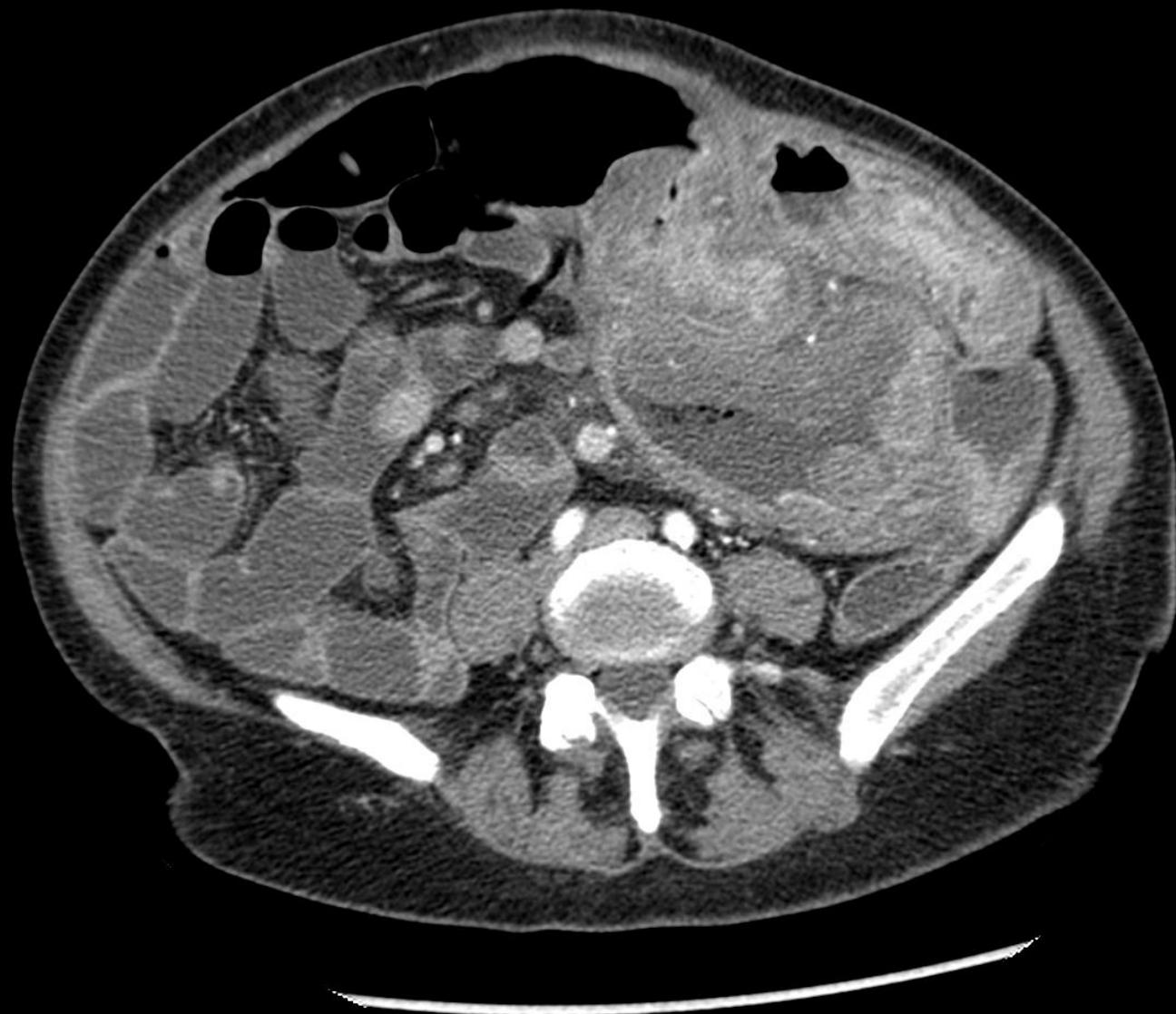


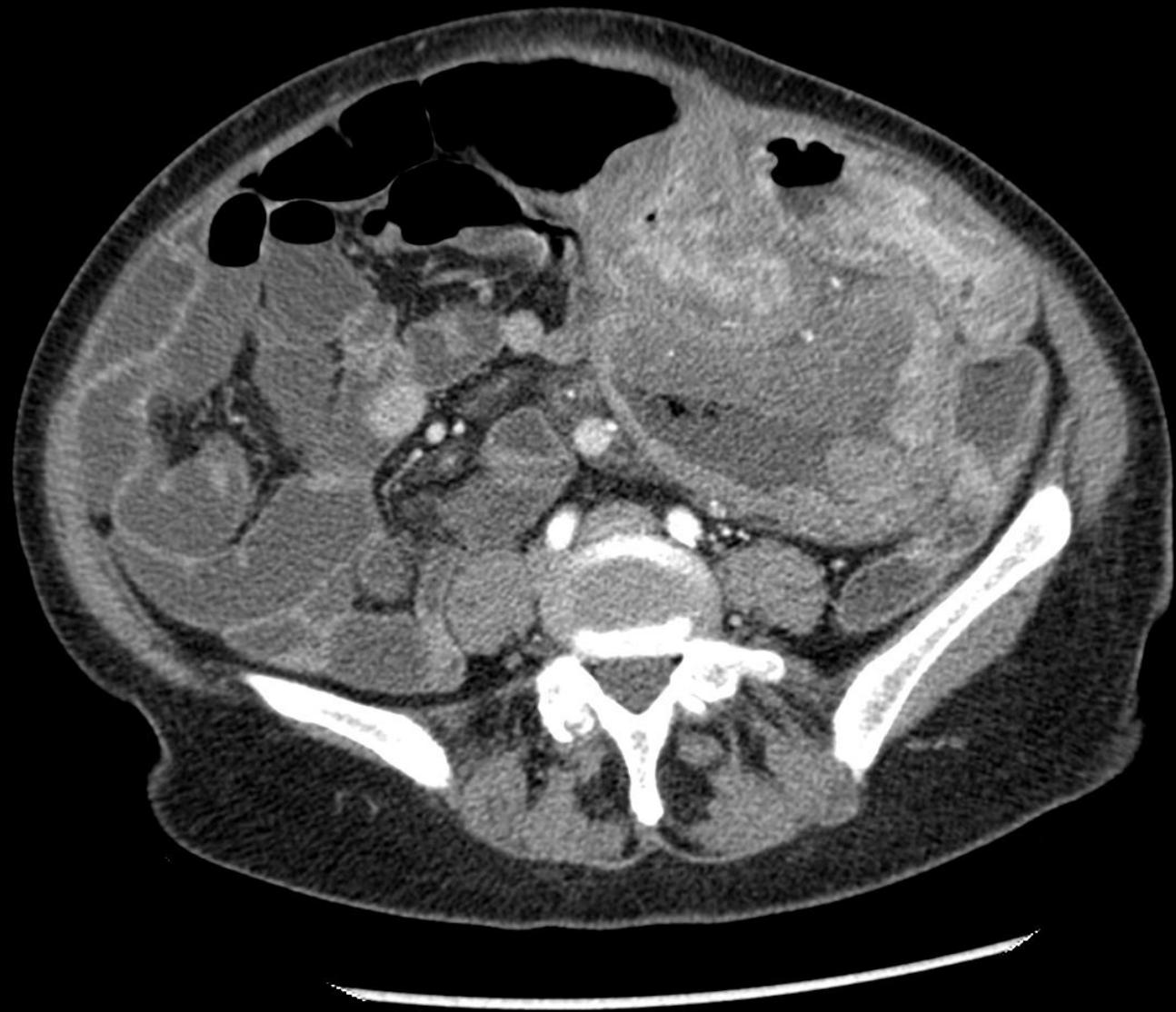


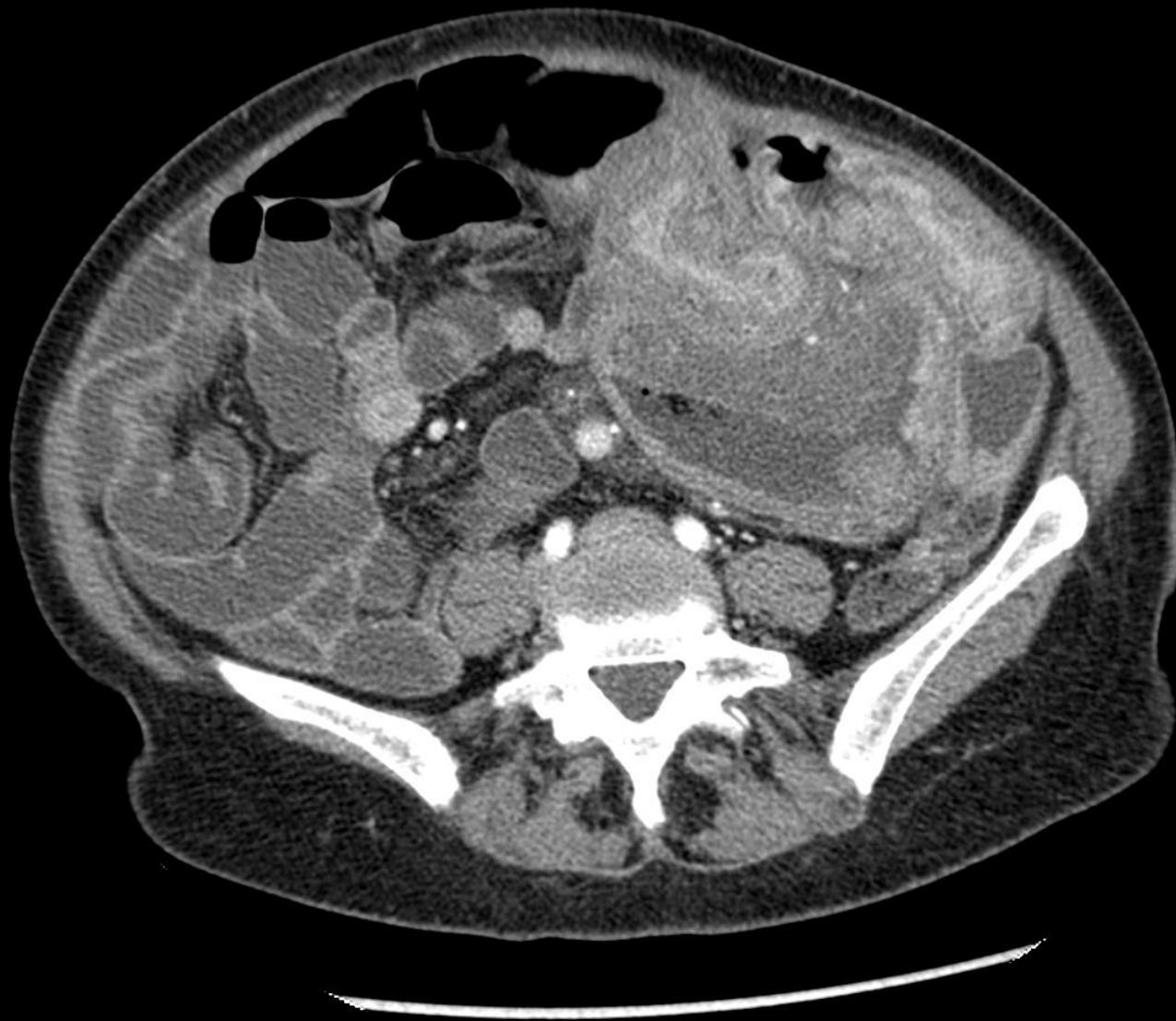














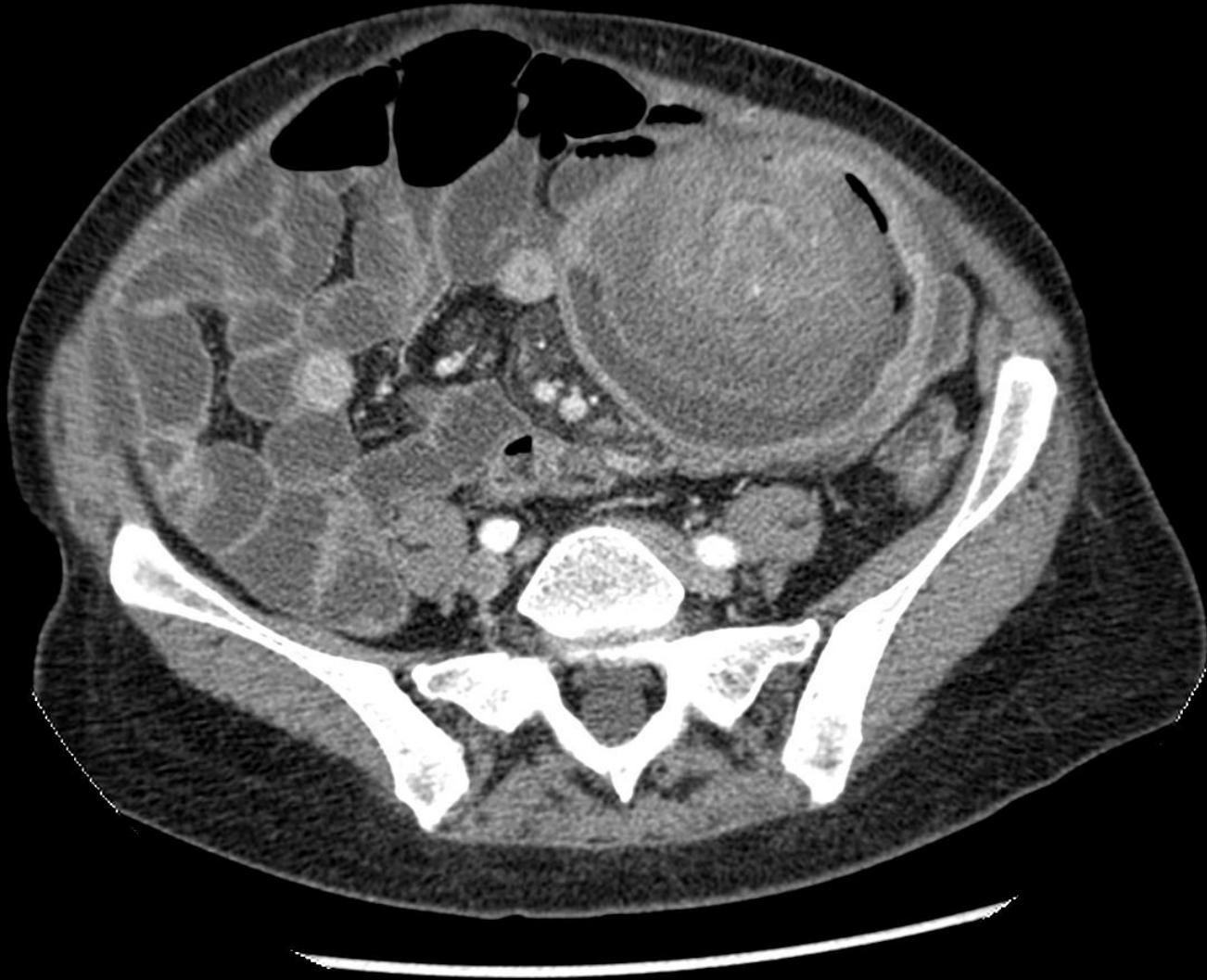


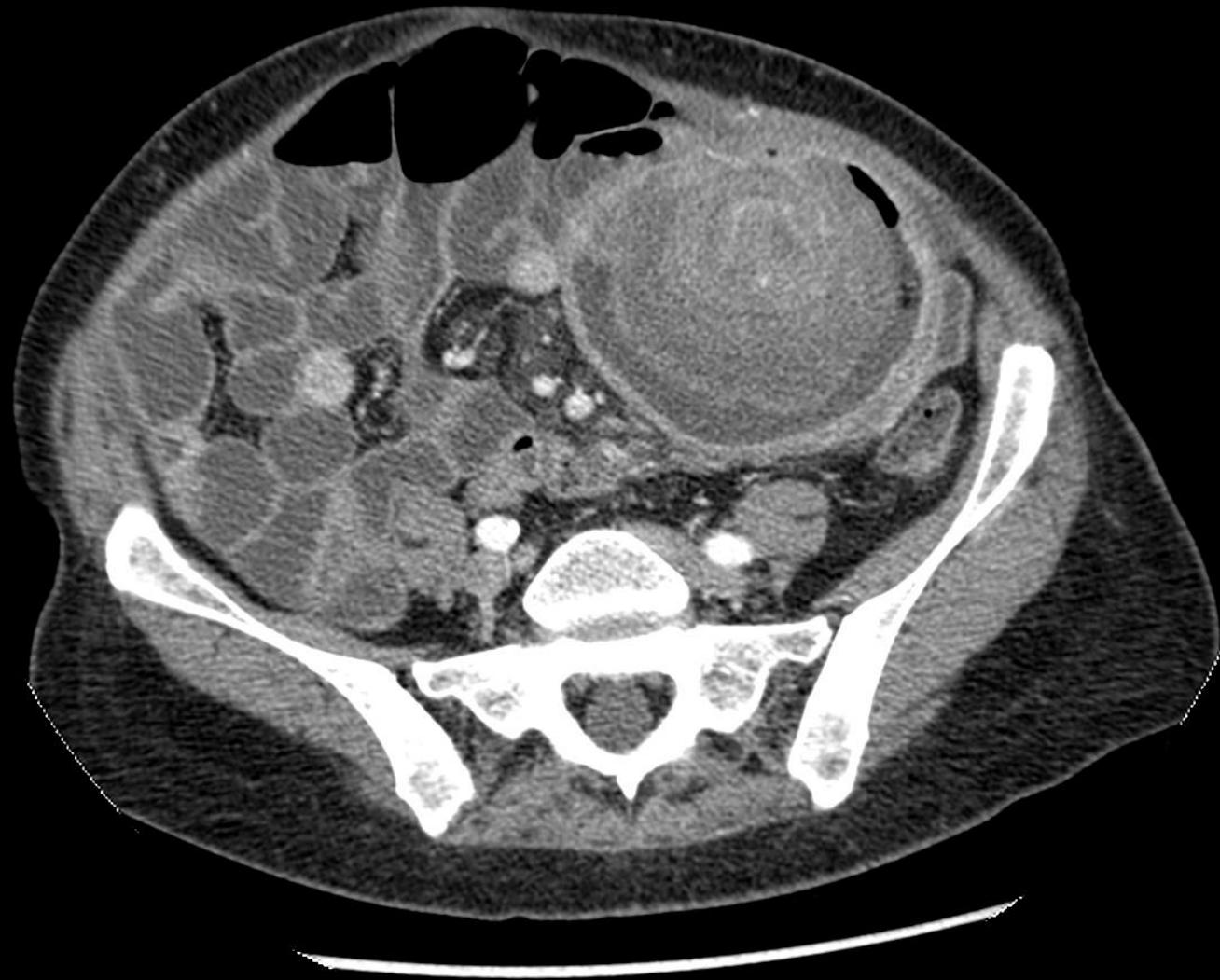












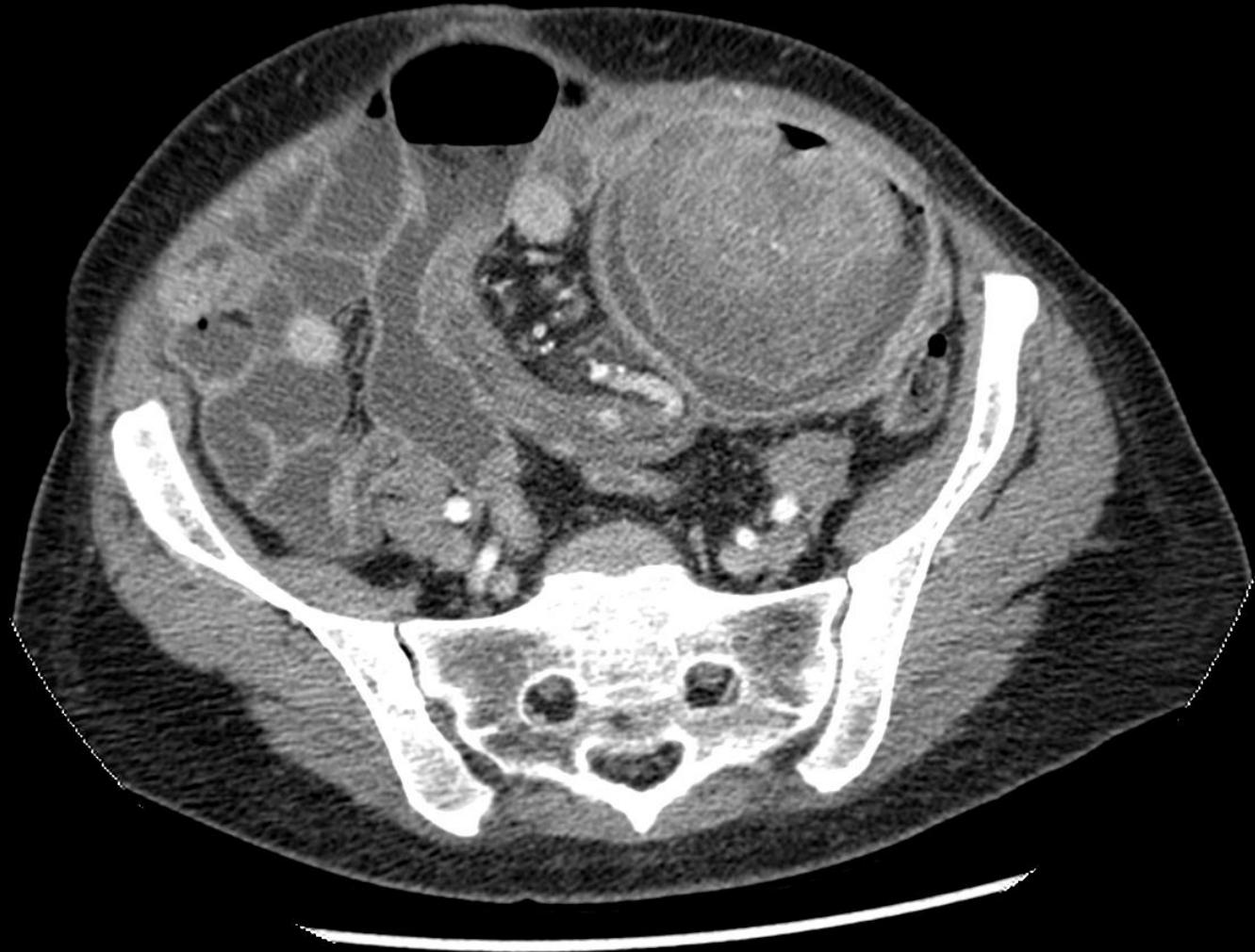






















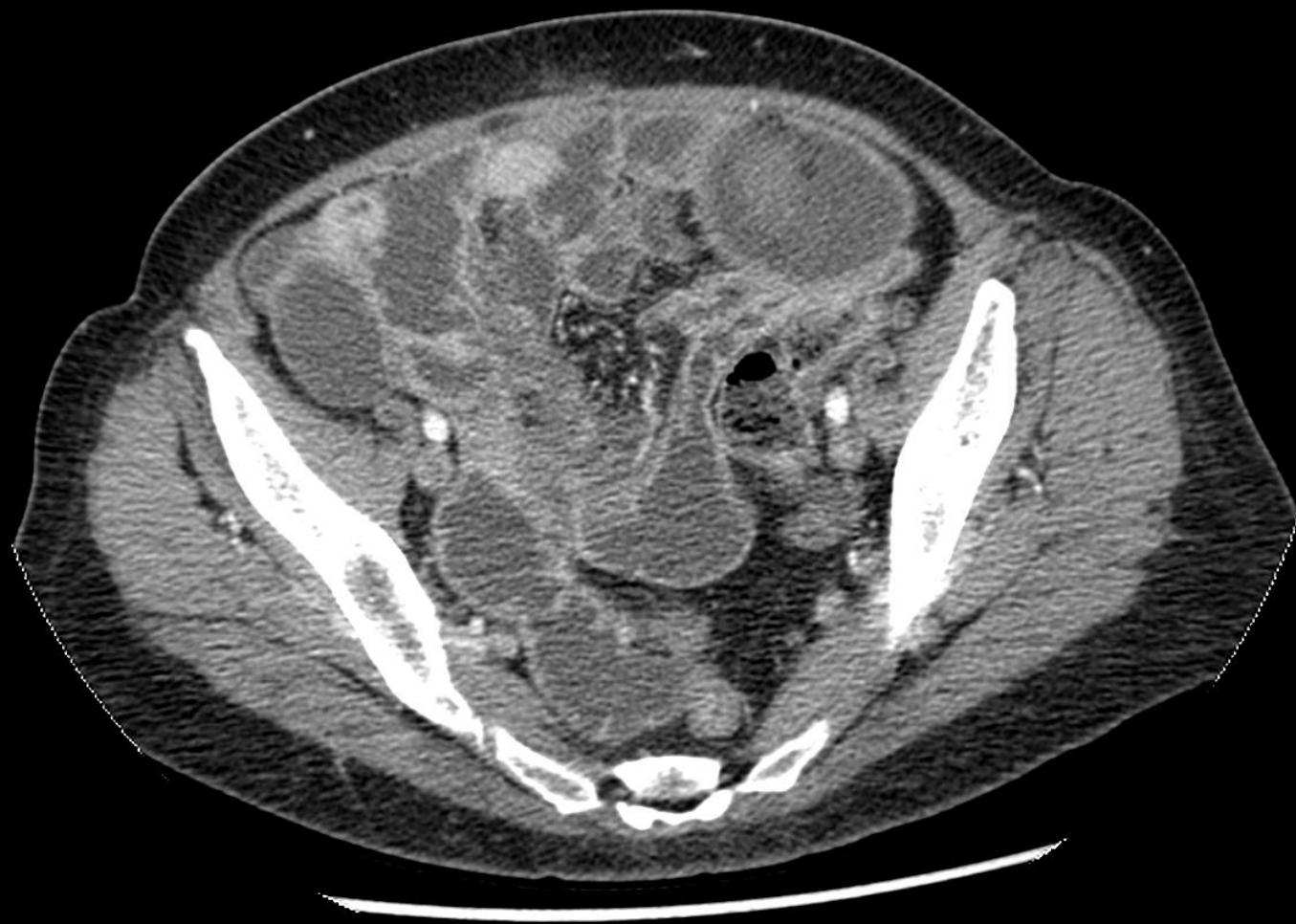






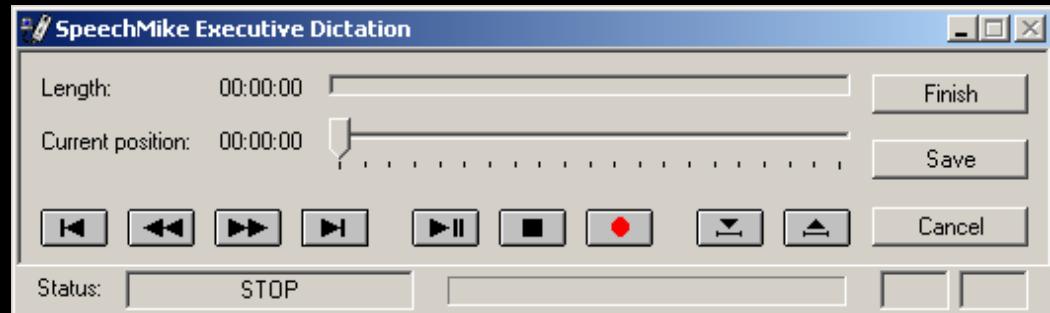












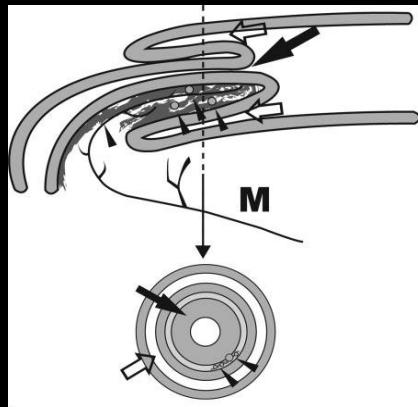
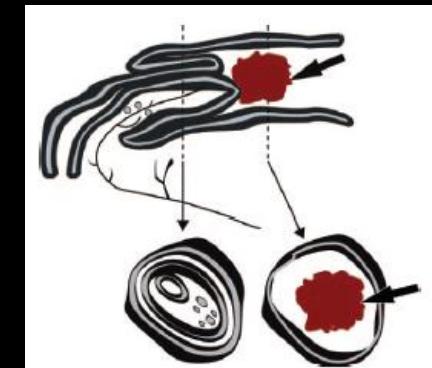


# Invaginação

- 5 % das invaginações ocorrem no adulto
- Localização, causa, ponto de partida
- *Coil spring*

Clinical Features of Adult Intestinal Intussusception

Feature	Intussusception without Lead Point	Intussusception with Lead Point
Duration	Transient, spontaneously resolving	Persistent or recurrent
Prevalence	Rare	Rare
Bowel obstruction	Often not present	Often present
Treatment	Unnecessary	Surgery required



Causes of Adult Intestinal Lead Point Intussusception

Nature of Cause	Location	
	Small Bowel	Large Bowel
Benign	Lipoma, adenomatous polyp, Meckel diverticulum	Lipoma, adenomatous polyp
Malignant	Metastasis, lymphoma, adenocarcinoma	Adenocarcinoma, lymphoma, metastasis
Idiopathic	Postoperative adhesion, motility disorder	Postoperative adhesion, motility disorder



# Invaginação

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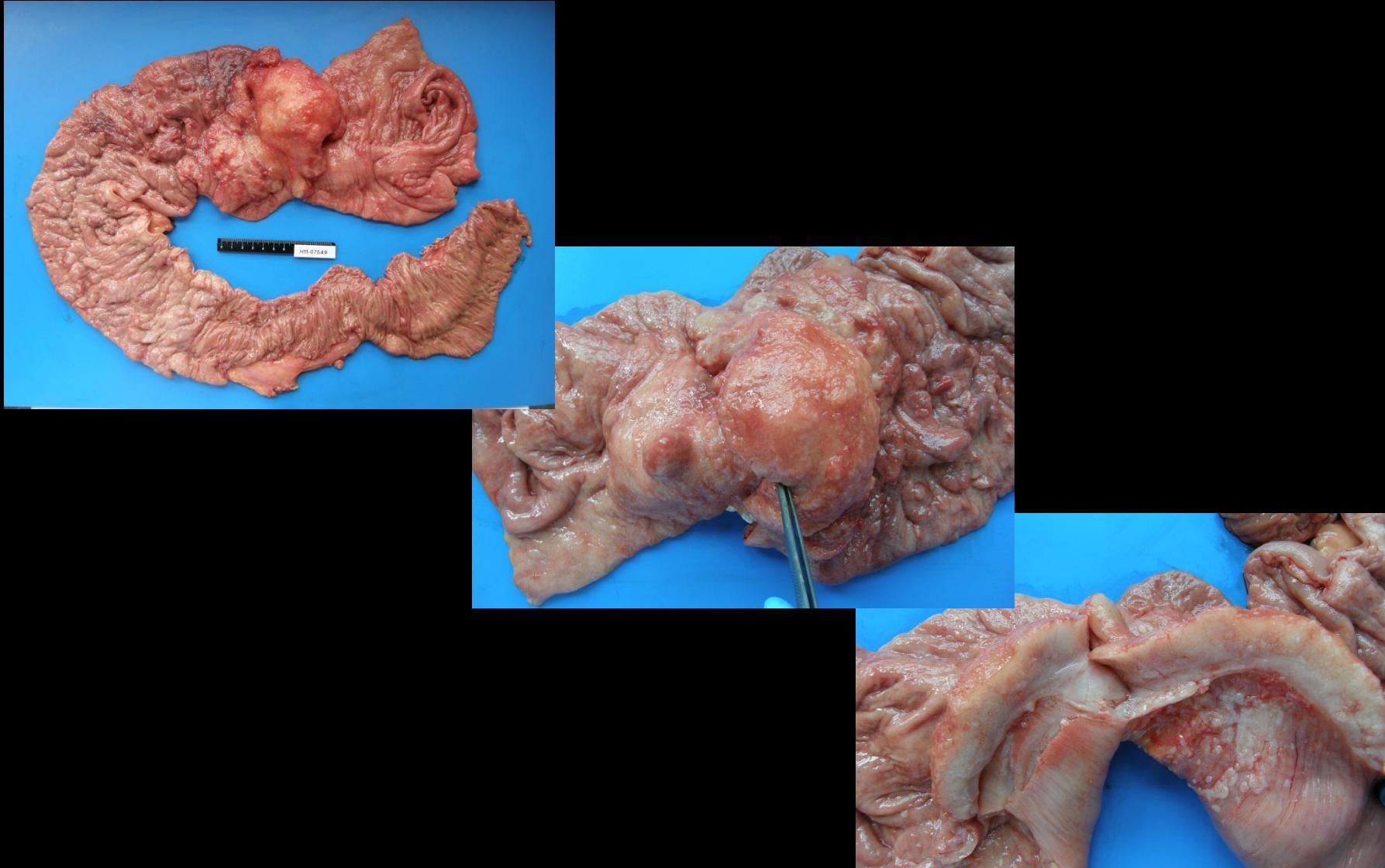


# Trânsito Intestino delgado (2011)





# Peça cirúrgica





# Anatomia Patológica

## Diagnóstico Histopatológico

Ganglioneurofibromatose difusa e nodular da parede do jejuno e neurofibromas com padrão plexiforme do mesentério.

Três adenocarcinomas invasores síncrones, dois superficiais, bem diferenciados (G1) com padrão tubular e um mucinoso que infiltra toda a parede, atinge o mesentério, tem embolização veno-linfática e doença metastática em 5, de 13 gânglios linfáticos regionais. Adenoma tubular séssil - neoplasia intra-epitelial de baixo grau.

pTNM - T3 N2 Mx.

AJCC - Estadio IIIB (se M0).

Tumor residual - R0.