Syndrome de Matthew-Wood

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Matthew-Wood Syndrome:

Report of Two New Cases Supporting Autosomal Recessive Inheritance and Exclusion of *FGF10* and *FGFR2*

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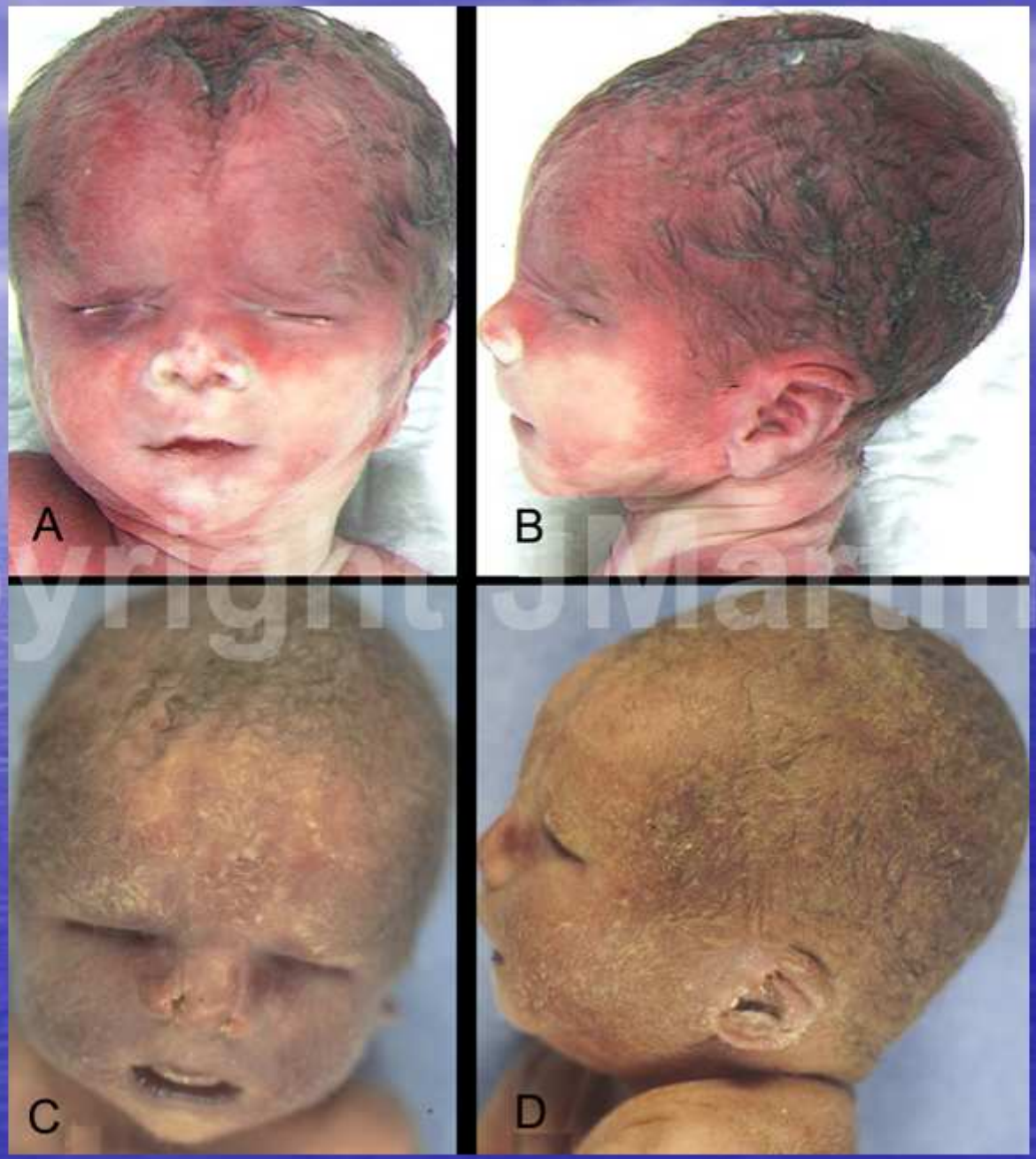


TABLE I. Comparison of clinical findings in Matthew-Wood Syndrome reported in the literature

Finding	Case 1	Case 2	Berkenstadt and al.	Spear and al.	Engellener and al.	Seller and al (2 sibs)	Steiner and al (2 sporadic cases)	Priolo and al.
Microphthalmia/ Anophtalmia	Bilateral	Bilateral	Bilateral	Bilateral	Unilateral	Bilateral (2/2)	Bilateral (2/2)	Bilateral
Pulmonary malformation	Bilateral agenesis of pulmonary artery branches	Bilateral agenesis	Unilateral agenesis	Bilateral agenesis	Bilateral agenesis	Bilateral agenesis of upper lobes (1/2) Bilateral hypoplasia (1/2)	No lung anomaly (1/2) Bilateral broncho-pulmonary dysplasia (1/2)	Bilateral hypoplasia
Diaphragmatic defect	Bilateral (eventration)	Bilateral (eventration)	Unilateral (hernia)	Unilateral (eventration)	Unilateral (inverted)	No (2/2)	Bilateral (eventration) (2/2)	Unilateral (hernia)
Cardiac malformation	no	Pulmonary artery agenesis, ventricular septal defect	no	Pulmonary artery agenesis, ventricular septal defect	Moderate dilatation of right atrium, closed narrow ductus	Single ventricle, hypoplastic left atrium, enlarged pulmonary trunk	Ostium primum ASD, right ventricular hypertrophy (1/2)	No
Facial Dysmorphia	Short palpebral fissures, short nose, retrognathia, large ears	Short palpebral fissures, long philtrum, large ears,	not documented	not documented	not documented	Prominent nose, short upper lip, micrognathia with cleft palate, Low-set ears (1/2)	Short palpebral fissures, Prominent nose with antevarted nares	
Other	Duodenal stenosis, hypoplastic pancreatic structures	Duodenal stenosis, polylobed spleen, agenesis of pancreas,	-	-	Renal dysplasia	Hypoplastic Spleen and uterus	Cryptorchidism (1/2)	malrotation of the left kidney
Growth retardation	+	+	+	+	+	+	+ (1/2)	+
Polyhydramnios	+	+	+	-	+	-	+ (1/2)	+
Karyotype	normal	normal	normal	normal	normal	normal	normal (2/2)	normal
Consanguinity	yes 3 sibs	yes	no	no	no	no 2 sibs	no (2/2)	no

Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene *STRA6*

Christelle Golzio, Jelena Martinovic-Bouriel, Sophie Thomas, Soumaya Mougou-Zrelli, Bettina Grattagliano-Bessières, Maryse Bonnière, Sophie Delahaye, Arnold Munnich, Férechté Encha-Razavi, Stanislas Lyonnet, Michel Vekemans, Tania Attié-Bitach, and Heather C. Etchevers

The American Journal of Human Genetics Volume 80 June 2007

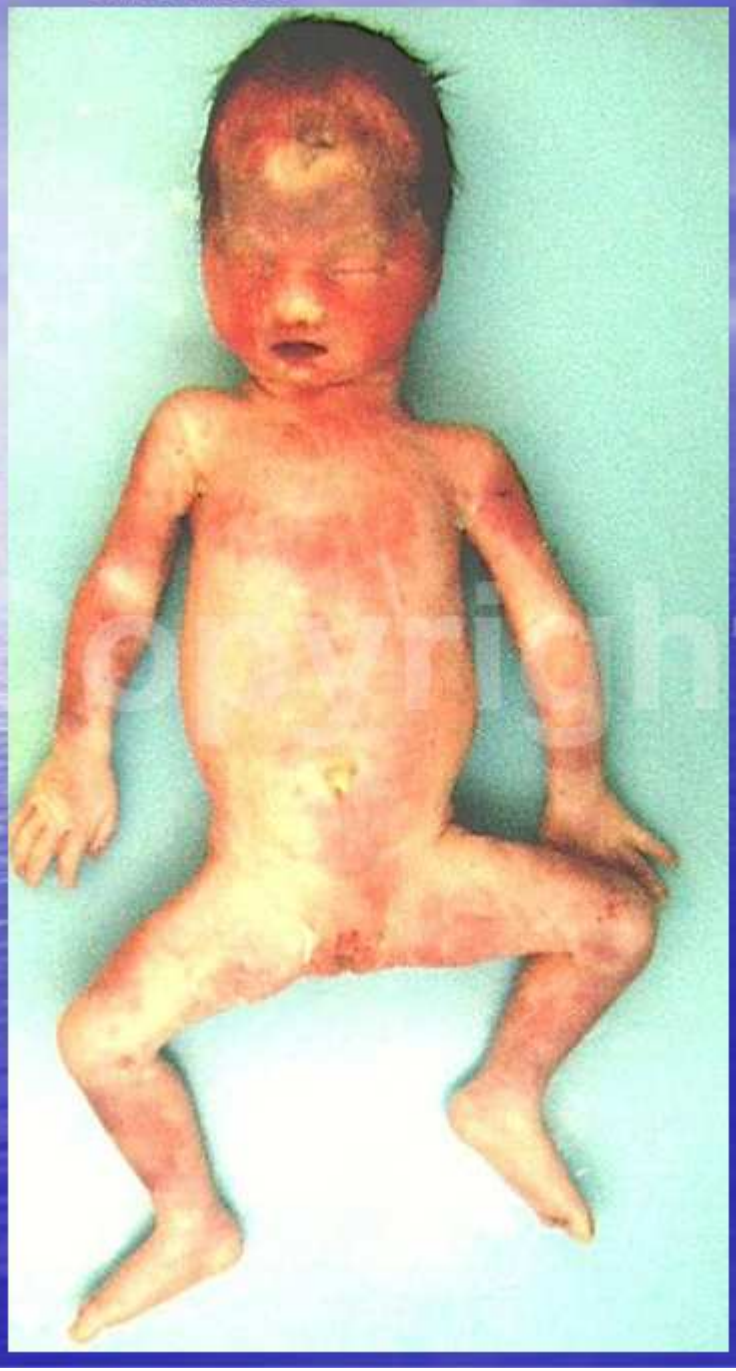
Mutations in *STRA6* cause a broad spectrum of malformations including anophthalmia, congenital heart defects, diaphragmatic hernia, alveolar capillary dysplasia, lung hypoplasia and mental retardation

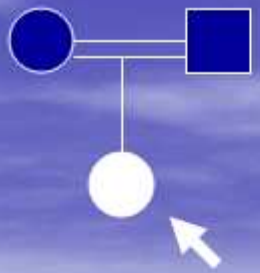
Running title: ***STRA6* mutations in human malformations**

Francesca Pasutto[1], Heinrich Sticht[2], Gerhard Hammersen[3], Gabriele Gillessen-Kaesbach[4], David R FitzPatrick[5], Gudrun Nürnberg[6,7], Heidemarie Schirmer-Zimmermann[3], John L Tolmie[8], David Chitayat[9], Gunnar Houge[10], Lorena Fernández-Martínez[1], Sarah Keating[11], Geert Mortier[12], Raoul CM Hennekam[13,14], Axel von der Wense[15], Anne Slavotinek[16], Peter Meinecke[17], Pierre Bitoun[18], Christian Becker[6,7], Peter Nürnberg[6,19], André Reis[1] and Anita Rauch[1]


C
D
E
F
G
200 µm

NAM...





Mère 1G 33ans

ETG :

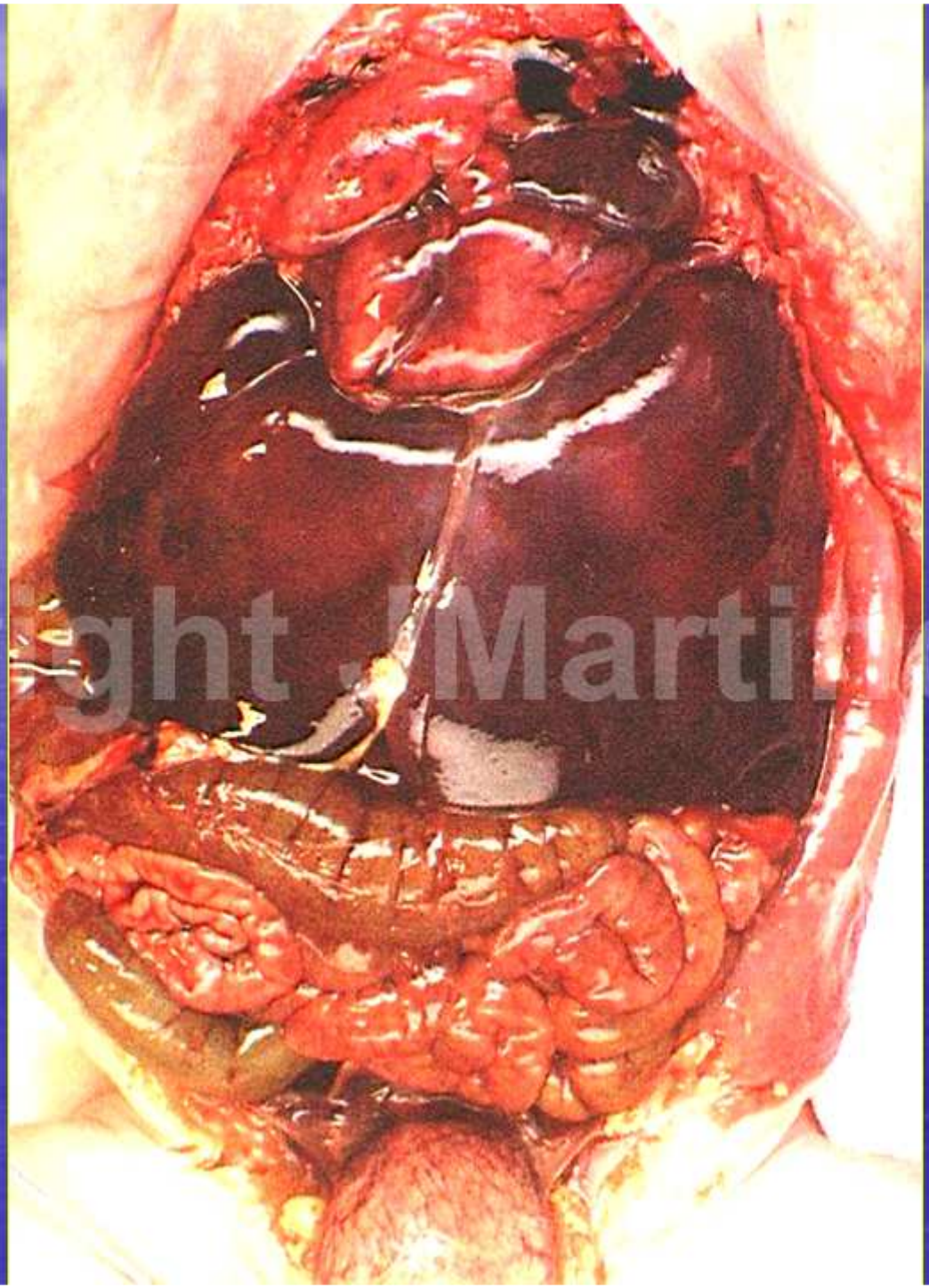
* hydramnios
* microphtalmie
* corps calleux au 10°p
* CIA
* reins: mauvaise diff. cortico-médullaire
* mégavessie

caryotype : normal

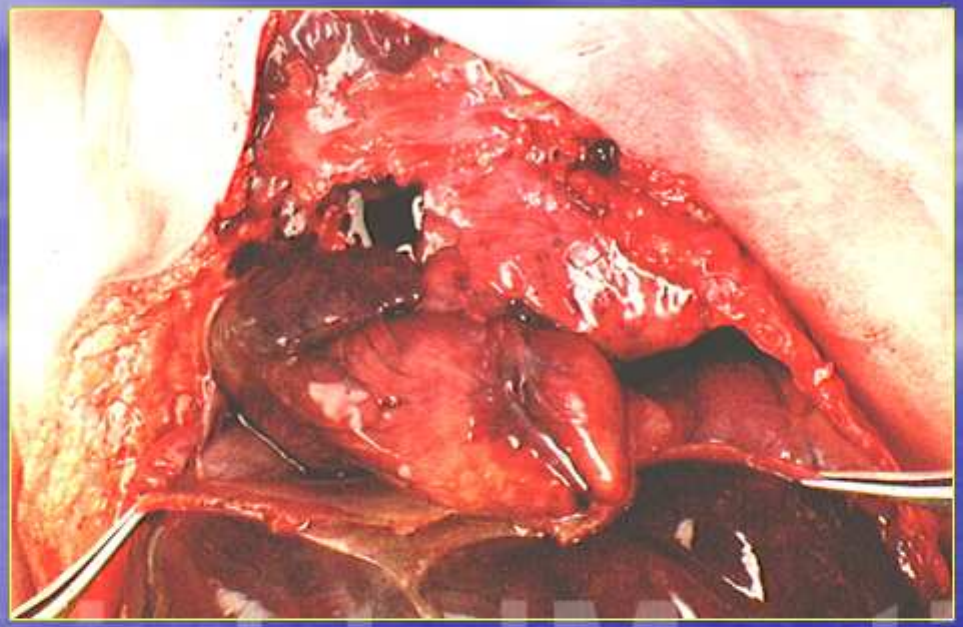
⟶ IMG à 35,6SA

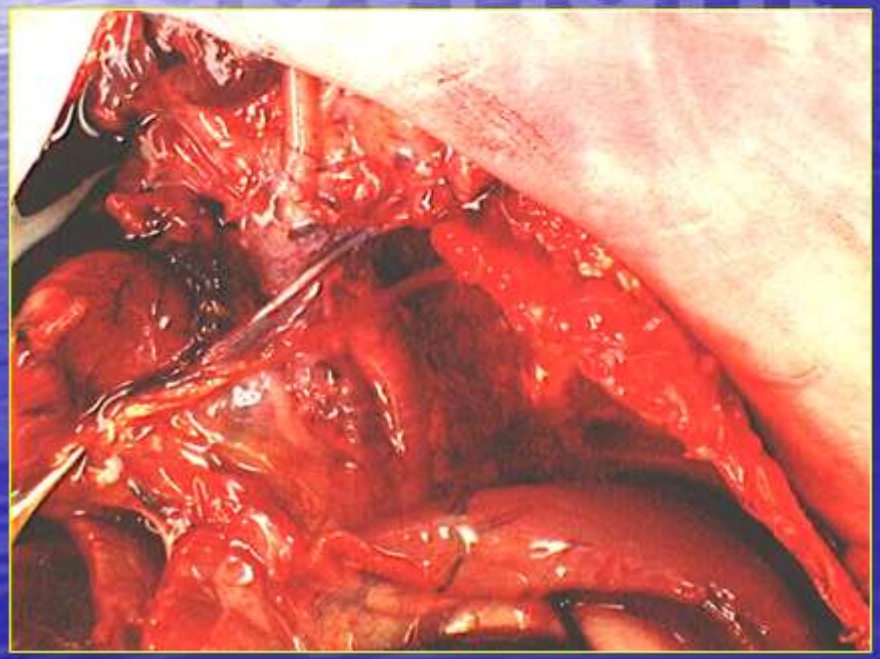
Dysmorphie+++

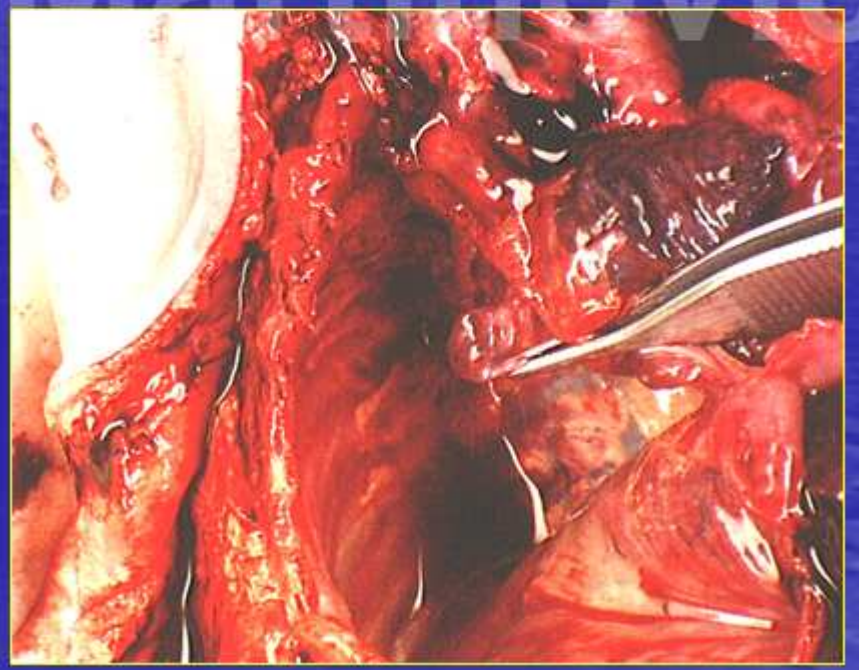
Evéntration
diaphragmatique

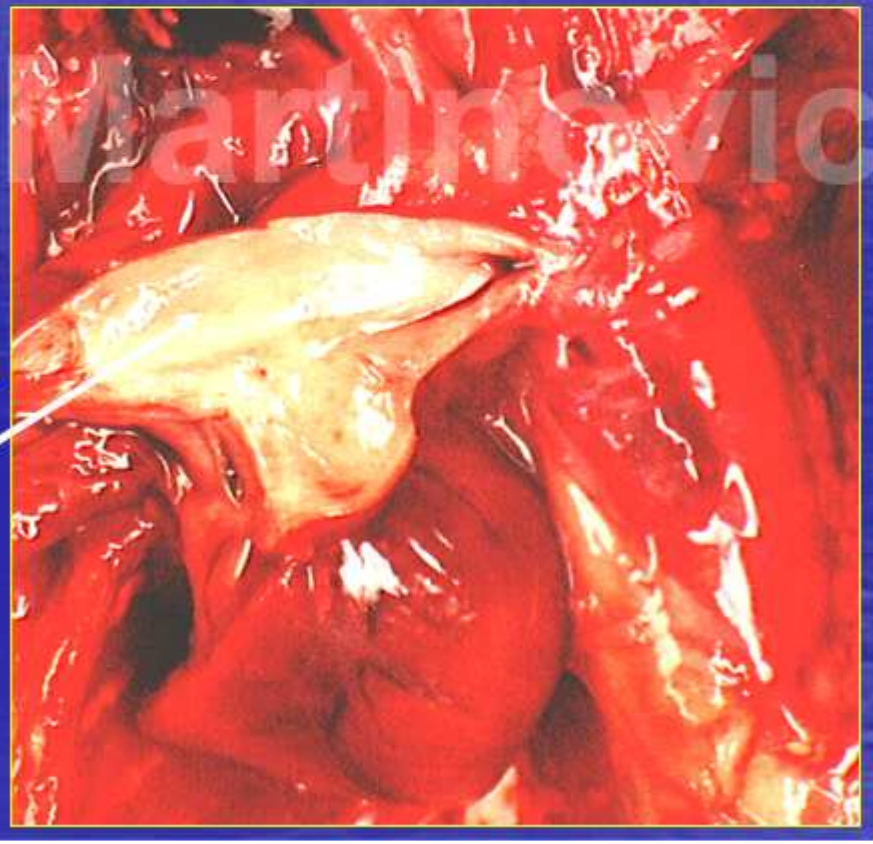


Agénésie pulm.
bilatérale





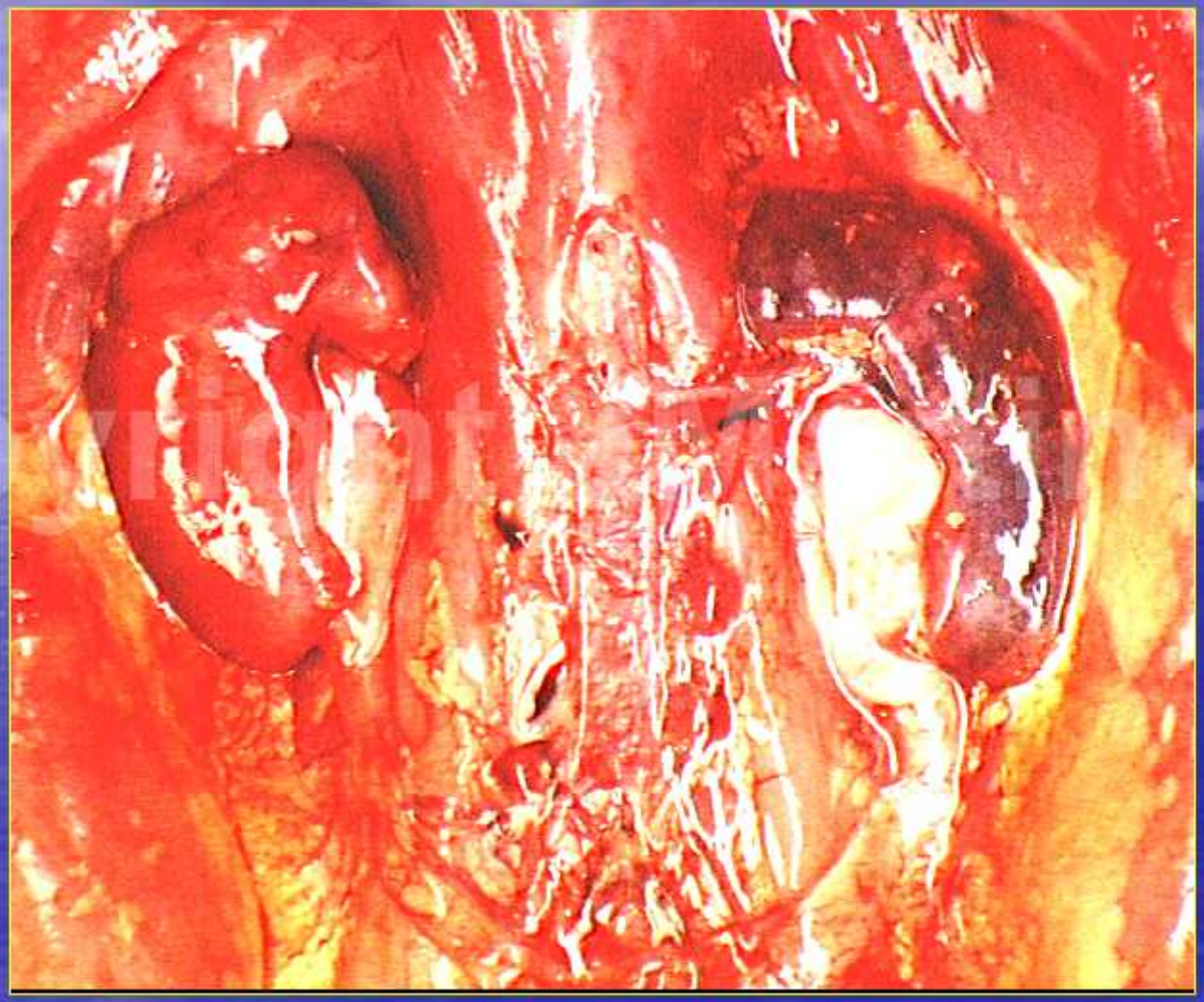




Agénésie des branches AP

Reins hypoplasiques avec dilatations pyéliques

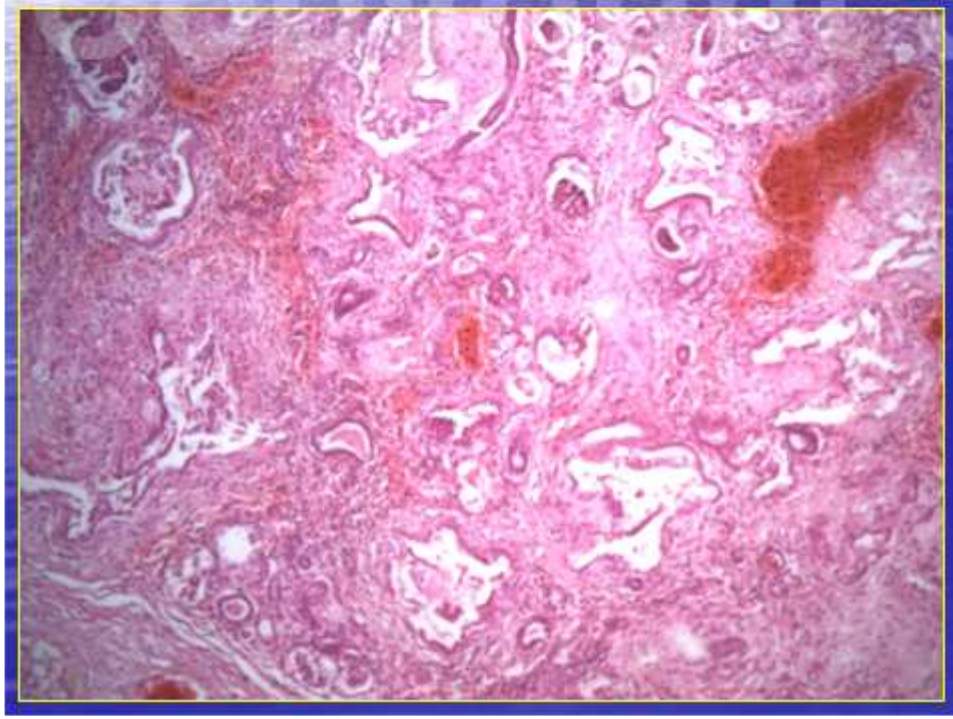
Dt : 5,2g

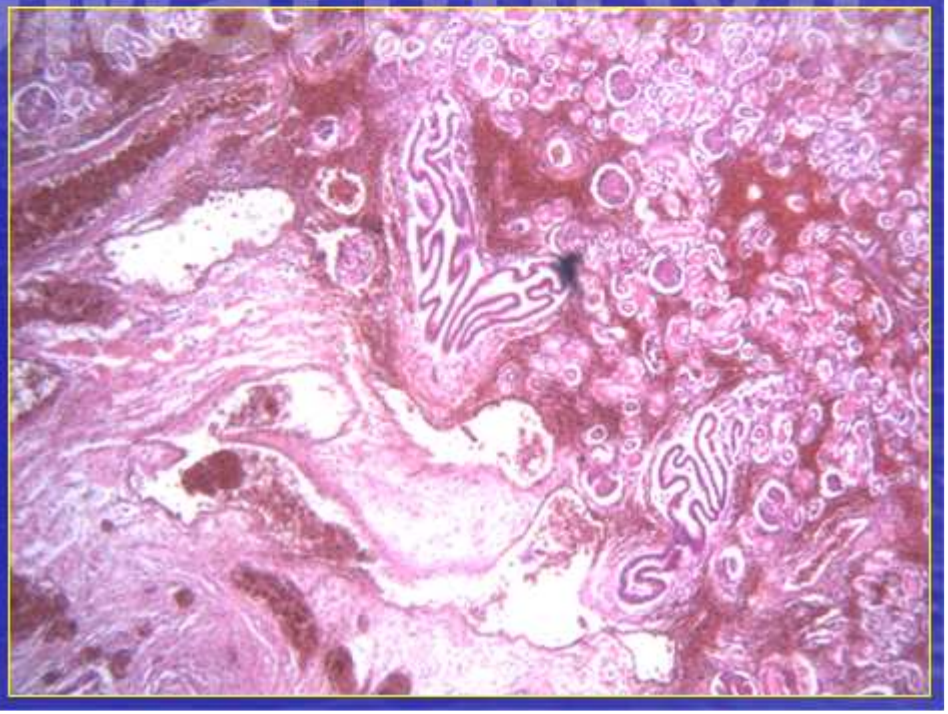


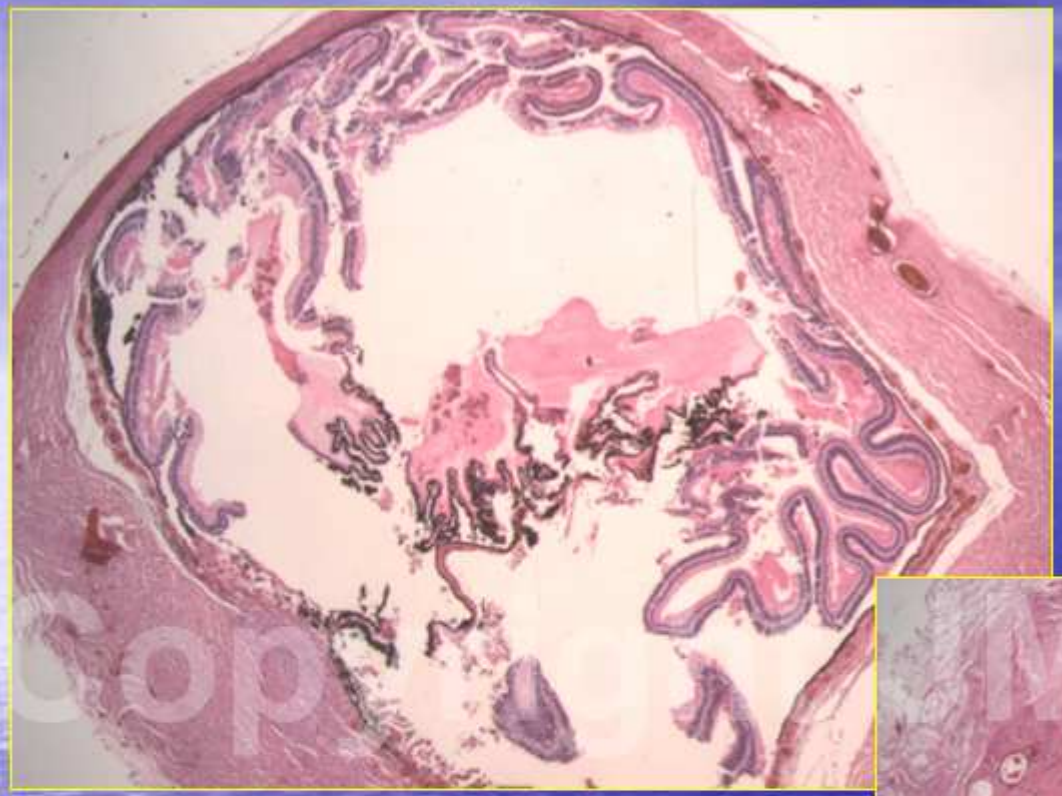
G : 5,1g

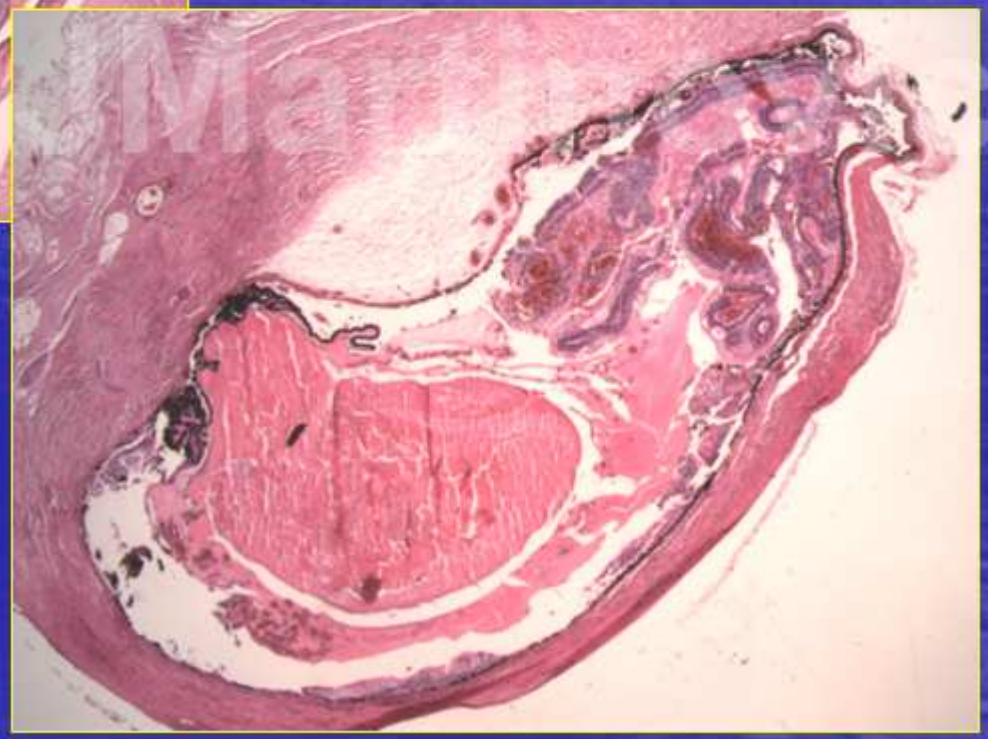
Nl poids combiné: 20±4g

Dysplasie
rénale bilat.









Microphtalmie bilat.
avec dysplasie rétinienne

Dg : Syndrome de Matthew-Wood

STRA6, RARB negatives…

Dg : Syndrome de Matthew-Wood

STRA6, RARB negatives

- Mutations HZ dans un nouveau gène
- Souris KO :anomalies du développement pulmonaire sévère

- Appel à d'autres familles/foetus
- Phenotype pulmonaire+++

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