NOT JUST ANOTHER CRYING BABY

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CASE

- PMA 39w1d, BW 3270g
- Age 14d
  - constant crying and agitation since birth
  - fever 38.1°C

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- PMA 39w1d, BW 3270g
- Age 14d
  - constant **crying** and agitation since birth
  - fever 38.1°C
  - always **hungry**, no effect of dietary changes
  - normal pattern of defecation
  - diapers from urinating still **very full**
  - familial history: multiple allergies, GERD

CASE

- Clinical exam

CASE

- Clinical exam
  - 3360g
  - **marbled skin**
  - agitated and **hungry** but alert, normatonic fontanel
  - systolic murmur left sternal border, normal femoral pulses
  - bloated abdomen, non tender

CASE

- Technical examination

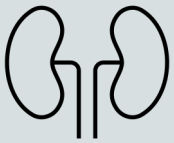
- blood sample

leucocytose	13.22	x10³/µl	7.30-16.60	
erythrocyten	4.09	x10e6/mm³	3.00-5.40	
hemoglobine	14.6	g/dL	11.0-18.0	
Hematocriet	42.5	%	35.0-55.0	
MCV	103.9	fL	85.0-120.0	
MCH	35.7	pg	28.0-40.0	
MCHC	34.4	g/dl	31.6-35.1	
Red blood cell distribution width	15.2	%	11.4-13.6	^
	RDW = een maat voor de variatie in het volume van de RBC			
thrombocyten	555	x10^3/µL	162-351	^
neutrofielen	30.7	%	14.0-51.0	
neutrofilie abs	4.06	x10^3/µL	1.00-8.50	
natrium	157.5	mmol/L	136.0-145.0	^
kalium	5.2	mmol/l	3.5-5.1	^
chloride	120.4	mmol/L	98.0-107.0	^
bicarbonaat	24.9	mmol/l	22.0-29.0	
calcium (mmol/L)	2.79	mmol/L	2.25-2.75	^
fosfor (mmol/L)	2.10	mmol/l	1.25-2.25	
creatinine	0.49	mg/dl	0.31-0.88	
AST	35	U/L	10-50	
ALT	28	U/L	10-50	
CK	158	U/L	<190	
CRP (mg/L)	<0.6	mg/L	0.0-5.0	
totaal eiwit (g/L)	59.8	g/l	51.0-73.0	

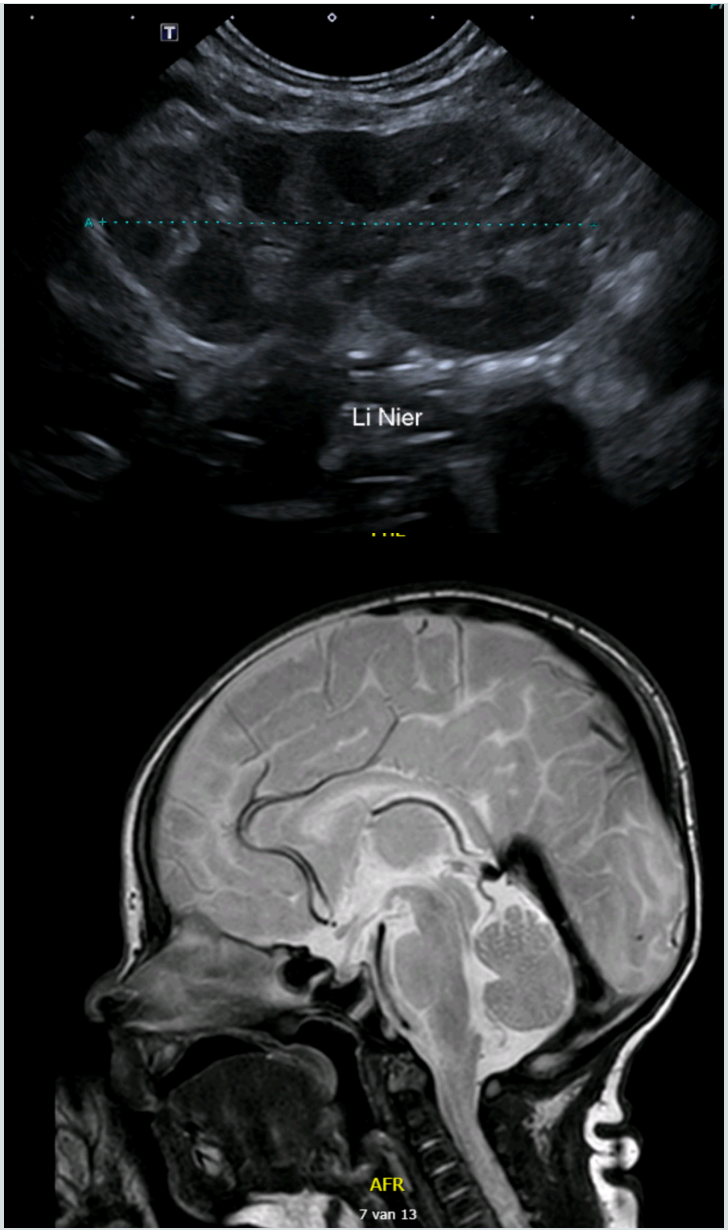
- urine sample

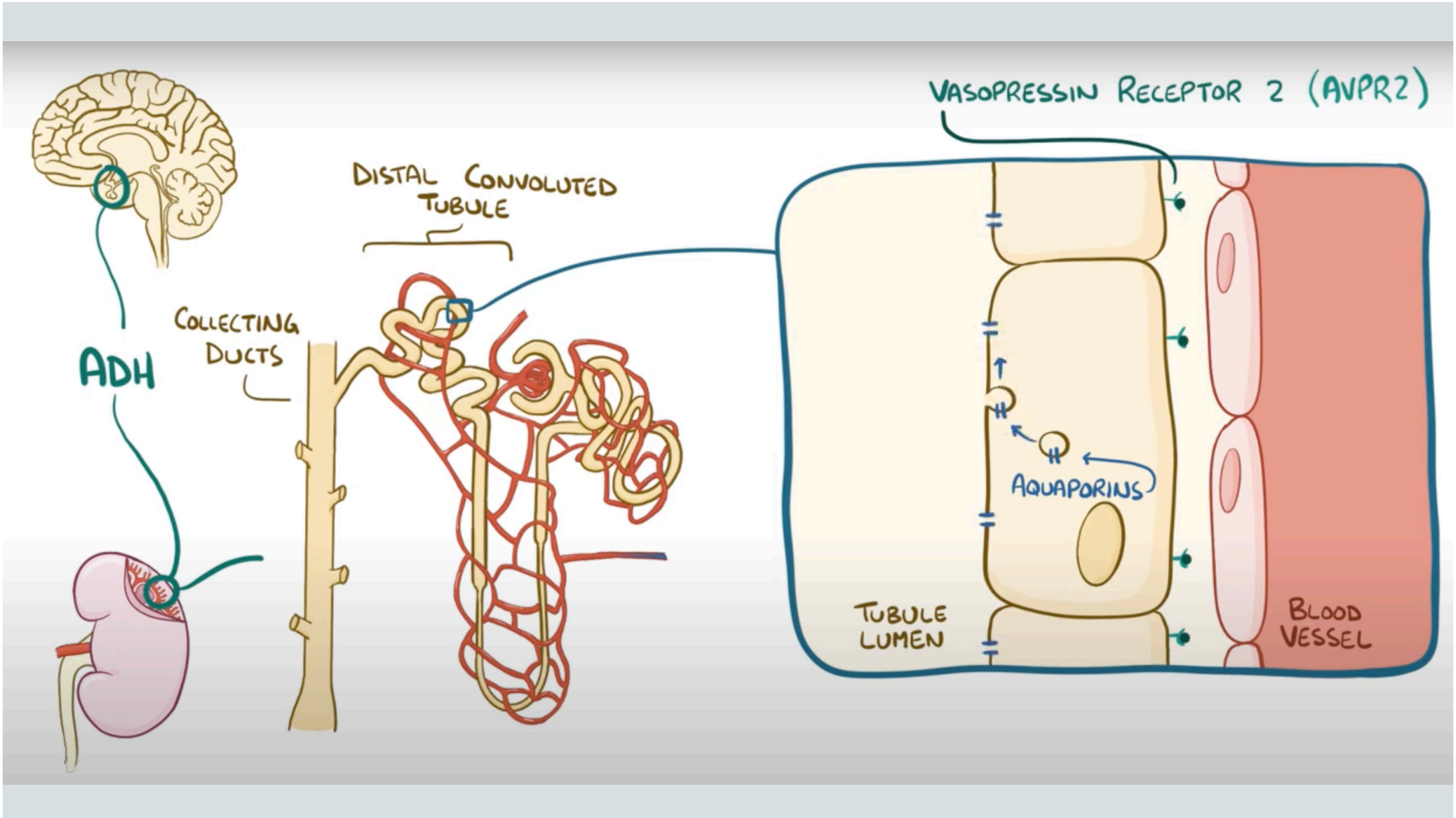
Erythrocyten	8.3	/µl	<25	
Leukocyten	1.8	/µL	<25	
hyaliene cilinders	0.14	/µL	<3.0	
pathologische cilinders	0.28	/µL	<1.0	
Totaal epitheelcellen	0.3	/µL	<25	
plaatepitheel	0.3	/µl	<25	
natrium	21.3	mmol/l		
kalium	27.4	mmol/l		
osmolaliteit	149	mosmol/kg	250-950	❯
creatinine	81	mg/l	400-2780	❯

FENa 1.06%



- abdominal ultrasound
  - normal aspect of kidneys and bladder
- cranial ultrasound
  - normal
- aEEG
  - normal
- brain MRI
  - normal pituitary gland




VASOPRESSIN RECEPTOR 2 (AVPR2)
DISTAL CONVOLUTED TUBULE
COLLECTING DUCTS
ADH
AQUAPORINS
TUBULE LUMEN
BLOOD VESSEL

CONGENITAL NEPHROGENIC DIABETES INSIPIDUS

- Genetic disease
  - AVPR2 (90%)
  - AQP2 (10%)
- X-linked
  - male: severe dehydration, hyperNa, hyperthermia, mental and physical retardation, kidney failure
  - female: variable, due to skewed inactivation x chromosome

hemizygous mutation c.541C>T (p.Arg181Cys) in the AVPR2 gene

TREATMENT

- low salt, low protein diet
- thiazide diuretics
- NSAIDs

REFERENCES

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Images from osmosis.com