

Online Resource 1. Biochemical Parameters in the CSHS cohort and published CSHS reports

	<b>Subject (CSHS# or author, age (years), sex)</b>	<b>Blood phosphate</b>	<b>Blood FGF23*</b>	<b>Alkaline phosphatase</b>	<b>25-OH- Vitamin D</b>	<b>1,25-OH<sub>2</sub>- Vitamin D</b>	<b>TRP or TmP/GFR</b>	<b>Blood calcium</b>	<b>PTH</b>	<b>Primary renal tubulopathy</b>	<b>Ref.</b>
Subjects from the CSHS cohort	CSHS101, 6/F	2.6 mg/dl (4-5.4)	445 pg/ml (10-50)	308 U/L (96-297)	29 ng/ml (33-100)	8 pg/ml (24-86) ↓	TRP 72%	2.4 mmol/L (2.15- 2.55)	15.9 pg/ml (15-65)	No	N/A
	CSHS102, 12/F	2.3 mg/dl (3.3-5.4)	279 RU/ml (<150)	366 U/L (70-280)	24ng/ml (33-100)	NR	LOW	9.7 mg/dl (8.8-10.8)	47 pg/ml (8.5-72.5)	No	N/A
	CSHS104, 3/F	1.5 mg/dl (3.9-6.5)	795 RU/ml (<150)	1085 U/L (108-317)	17 ng/ml (10-80)	10 pg/ml (24-86)	TRP 80%	2.1 mmol/L (2.15- 2.55)	67 pg/ml (15-65)	No	N/A
	CSHS105, 16/M	2.6 mg/dl (2.8-4.6)	74 pg/ml (10-50)	283 U/L (52-171)	27 ng/ml (33-100)	69 pg/ml (24-86)	TRP 84%	2.2 mmol/L (2.15- 2.55)	47 pg/ml (15-65)	No	N/A
	CSHS106, 13/F	1.3 mg/dl (3.3-5.4)	176 pg/ml (10-50)	1186 U/L (50-162)	19 ng/ml (33-100)	37 pg/ml (24-86)	TRP 79%	2.3 mmol/L (2.15- 2.55)	52 pg/ml (15-65)	No	N/A
Subjects from the literature in whom FGF23 was assessed	Narazaki et al. 5/F	2.1mg/dL (3.8-5.8)	123 pg/ml (10-50)	1935 U/L (330-880)	16 pg/ml	32 pg/ml (20-40)	TmP/GFR 1.9mg/dl (4.7-5.6)	9.9 mg/dL	61 pg/mL (15-65)	NR	[1]
	Sethi et al. 5/M	2.1 mg/dl (3.5-5.5)	1265 pg/ml (10-50)	NR	55nMol/L (>50)	NR	TmP/GFR 1.5mg/dl (2.9-4.6 mg/dl)	10.9mg/dl (9-11)	338 pg/ml (10-60)	No	[2]
	Lim et al. CSHS 103 15/F	1.5mg/dl (3.3-5.4)	524 RU/ml (<150)	651 U/L (30- 120)	LOW	NR	LOW	9.3 mg/dl (9-10.5)	90 pg/ml (10-60)	No	[1]
	Hoffman et al. 16/M	1.9 mg/dl (3.5-5.2)	530 RU/ml (<150)	534 U/L	36 (10- 55ng/nl)	48 pg/ml (21-65)	TmP/GFR 1.6 mg/dl (3.2±0.3)	9.8 (8.5- 10.2)	45 pg/ml (10-65)	No	[3]
	Aggarwal et al. 10/M	2.1 mg/dl (2.1-4.5)	171RU/ml (<150)	2685 U/L (240-840)	69ng/ml	NR	TRP 73%	8.7(8.1- 10.4)	127.7 pg/ml	No	[4]
	Avitan- Hersh et	1.9 mg/dl (3.6-5)	69pg/ml (10-50)	NR	7ng/ml (>30)	21 pg/ml (25-76)	TRP 96%	9.8 (8.5- 10.6)	62ng/L(15-65)	No	[5]

al. 14/M										
Heike et al. 19/M	2.2 mg/dl (4.3-5.7)	215 RU/ml (<150)	1103 U/L (133-347)	12 (13-67ng/ml)	19 (27-71 pg/ml)	TmP/GFR 1.5 mg/dl (2.5-4.2)	9.4 (9.4-10.6)	51 (11-47 pg/ml)	No	[6]

	Subject (author, age (years), sex)	Serum phosphate	Serum FGF23*	Alkaline phosphatase	25-OH-Vitamin D	1,25-OH <sub>2</sub> -Vitamin D	TRP or TmP/GFR	Serum calcium	PTH	Primary renal tubulopathy	Ref
<b>Subjects from the literature in whom FGF23 was not assessed</b>	Moreira et al. 1/F	1.9mg/dl (3-6.5)	NR	1192 U/L (54-280)	normal	NR	LOW	normal	normal	No	[7]
	Kishida et al. 7/F	2.1 mg/dl (4.5-6.5)	NR	270 U/I (<116)	NR	NR	LOW	9.8 mg/dl (8.5-10.5)	normal	No	[8]
	Chou et al. 6/M	1.5 mg/dl (3.7-5.6)	NR	1041 U/L (145-420)	8.94 ng/ml (9.7-41.7)	NR	TRP 80%	9.5 mg/dl (8.8-10.8)	94.6 pg/ml(10-65)	No	[9]
	Chou et al. 22/M	1.5 mg/dl (2.9-5.4)	NR	437 U/L (70-230)	NR	NR	TRP 91%	9.7mg/dl	35.3 (10-65)	No	[9]
	Olivares et al. 2.5/F	0.4mmol/L (0.9-1.75)	NR	350 U/L (37-200)	70 nmol/L (37-200)	21.6 pmol/L (35-105)	TRP 42%	9.2 mg/dl (8.8-10.8)	NR	No	[10]
	Ivker et al. 1/F	0.87 mmol/L (1.54-2.62)	NR	420 (U/L) (50-136)	normal	36 nmol/L (36-144)	TRP 68.5%	normal	28 ng/L (51-217)	NR	[11]
	Oranje et al. 4/M	0.7 mmol/L (1-1.8)	NR	453 U/L (80-225)	100umol/L (31-129)	46 nmol/L (40-140)	TRP 86%	normal	2.2 pmol/L (0.8-15)	No	[12]
	Tokatli et al. 5/M	1.7 mg/dl	NR	25 U/L	NR	NR	NR	10.6 mg/dl	NR	NR	[13]
	Stosiek et al. 20/M	2.4 mg/dl (low-normal)	NR	304 U/L (high)	21.4 ng/ml (N)	11pg/ml (10-16)	TRP 87%	9.2 mg/dl	5.3 pmol/L (normal)	NR	[14]
	O'Neill et al. 3/F	1.1 mmol/L	NR	600 U/L	NR	NR	NR	8.4 mg/dl	NR	NR	[15]
	Skokby et al. 1/M	0.54 mmol/L (1.24-1.84)	NR	1472 U/L (250-1000)	NR	91 pmol/L (24-158)	TRP 37%	normal	normal	No	[16]
	Carey et al. 7/M	2.6 mg/dl	NR	832 IU/L	34.4 ng/ml (15-80)	4.3 pg/ml (21-74)	TmP/GFR 1.58 mg/dl (4-5.9)	9.3 mg/dl	0.9 ng/ml (0.4-1.5)	No	[17]
Carey et al. 23/F	1.2 mg/dl	NR	306 IU/L	18 ng/ml (15-80)	8.7 pg/ml (19-50)	TmP/GFR 0.64 mg/dl (2.5-4.2)	8.8 mg/dl	0.8 ng/nl (0.4-1.5)	No	[17]	

	<b>Subject (author, age (years), sex)</b>	<b>Serum phosphate</b>	<b>Serum FGF23*</b>	<b>Alkaline phosphatase</b>	<b>25-OH- Vitamin D</b>	<b>1,25-OH<sub>2</sub>- Vitamin D</b>	<b>TRP or TmP/GFR</b>	<b>Serum calcium</b>	<b>PTH</b>	<b>Primary renal tubulopathy</b>	<b>Ref</b>
<b>Subjects from the literature in whom FGF23 was not assessed</b>	Sugarman et al. 12/F	2.5 mg/dl (3-5.5)	NR	210 U/L (30-130)	NR	NR	NR	8.9 mg/dl (9-11)	NR	moderate aminoaciduria	[18]
	Moorjani et al. 8/F	2.5 mg/dl	NR	350 IU/L	NR	NR	NR	9.5 mg/dl	NR	trace of amino acids	[19]
	Aschinberg et al. 12/M	1.3 mg/dl (2.5-5)	NR	313 IU/L (20-124)	NR	NR	TRP 35%	9.6 mg/dl (9-11.5)	523 pg/ml (275-675)	No	[20]
	Hosalkar et al. 7/M	1.1 mg/dl (2.5-5)	NR	340 IU (20-124)	18 ng/ml (15-80)	6 pg/ml (20-75)	TRP 40%	8.9 mg/dl (9-11.5)	0.8ng/ml (0.4-1.5)	NR	[21]
	Hosalkar et al. 6/M	1.3 mg/dl (2.5-5)	NR	330 IU (20-124)	12 ng/ml (15-80)	9 pg/ml (20-75)	TRP 35%	8.8 mg/dl(9-11.5)	0.9ng/ml (0.4-1.5)	NR	[21]
	Hosalkar et al. 3/M	1.8mg/dl (2.5-5)	NR	210 IU (20-124)	14 ng/ml (15-80)	11 pg/ml (20-75)	TRP 46%	9.1 mg/dl (9-11.5)	0.5ng/ml (0.4-1.5)	NR	[21]
	Rustin et al. 35/M	Low/normalized	NR	NR	NR	NR	NR	normal	normal	NR	[22]
	Sukkhoja et al. 4/F	0.6mmol/L (1.2-1.8)	NR	2877 (169-372 U/L)	61nmol/L (>50)	NR	TRP 75%	9.2mg/dl	12.5pmol/l (3.7-15.9)	NR	[23]
	Shahgholi et al. newborn/F	1.9mg/dl (3.5-6)	NR	3300 U/L (45-300)	normal	NR	NR	normal	NR	NR	[24]
	de Morais et al. 9/F	LOW	NR	1630 U/L (175-420)	normal	NR	NR	normal	normal	NR	[25]
	Feldelmann et al. 19/F	0.43 mmol/L (1.11-1.5)	NR	107 U/L (30-90)	86ng/ml (5-70)	26 pg/nl (15-20)	TRP 51%	normal	0.15ng/ml (<0.5)	normal	[26]
	Shieh et al. 9/M	1.3mg/dl	NR	NR	NR	NR	67%	normal	0.78ng/ml (0.22-0.66)	NR	[27]

	<b>Subject (author, age (years), sex)</b>	<b>Serum phosphate</b>	<b>Serum FGF23*</b>	<b>Alkaline phosphatase</b>	<b>25-OH- Vitamin D</b>	<b>1,25-OH<sub>2</sub>- Vitamin D</b>	<b>TRP or TmP/GFR</b>	<b>Serum calcium</b>	<b>PTH</b>	<b>Primary renal tubulopathy</b>	<b>Ref</b>
<b>Subjects from the literature in whom FGF23 was not assessed</b>	Zutt et al. 15- 52/F	low/ normalized	NR	high/ normalized	NR	NRA	low/ normalized	normal	NR	normal	[28]
	Saraswat et al. 22/M	2.4mg/dl (2.5-4)	NR	46 U/dl (5-14)	NR	NR	phosphaturia	7.8mg/dl (8-11)	normal	NR	[29]
	Bouthors 52/M	0.67mmol/l	NR	normal	NR	NR	76%	normal	normal	NR	[30]
	Gathwala et al. 6/M	2.6mg/dl	NR	1976 U/L	normal	NR	phosphate urinary excretion 700mg/d (100-300)	8.7mg/dl	normal	normal	[31]
	John et al. 7/M	1.8mg/dl	NR	982 U/L	NR	NR	TmP/GFR 0.5mg/dl	9.6mg/dl	NR	normal	[32]
	Klein et al. 2/M	2.5mg/dl	NR		32.6 ng/ml (normal)	21.6pg/ml (normal)	79%	normal	35pg/ml (normal)	NR	[33]
	Sanmanechai et al. 7/M	0.77mmol/L (1.19-1.8)	NR	630U/L(145- 420)			64%	2.22mmol/L (2.2-2.7)	8.38pmol/l (0.95-6.84)	normal	[34]
<b>Subjects with no reported mineral anomalies or no labs reported</b>	Cabanillas et al. 26/M	normal	normal	normal	NR	normal	NR	normal	normal	NR	[35]
	Pierini et al. 9/M	4.9mg/dl normal	NR	NR	NR	NR	NR	9.5mg/dl (normal)	NR	NR	[36]
	Kaplan et a. 14/F	NR	NR	NR	NR	NR	NR	normal	normal	NR	[37]
	Grun 13/M	normal	NR	NR	NR	NR	NR	normal	NR	NR	[38]

Abbreviations: NR: Not reported, TRP: renal tubular reabsorption of phosphate; TmP/GFR: Tubular maximum Phosphate Reabsorption per Glomerular Filtration

	Subject (CSHS# or author, age (years), sex)	Serum phosphate	Serum FGF23*	Alkaline phosphatase	25-OH- Vitamin D	Calcitriol	TRP or TmP/GFR	Serum calcium	PTH	Primary renal tubulopathy	Ref
<b>Subjects with no reported mineral anomalies or no labs reported</b>	Camacho et al. 18/M	NR	NR	544 U/L (30- 115)	NR	NR	NR	NR	NR	NR	[39]
	Bouwes 19/F	NR	NR	NR	NR	NR	NR	NR	NR	NR	[40]
	Vidaurre et al. x 3patients	NR	NR	NR	NR	NR	NR	NR	NR	NR	[41]
	Yu e tal. 9/M	NR	NR	NR	NR	NR	NR	NR	NR	NR	[42]

Normal ranges for serum phosphate in our cohort are adjusted to the age of the subject.

Serum FGF23 values are displayed as intact (pg/ml) or total value (intact plus C-terminal fragments) RU/ml.

Normal ranges for biochemical parameters in subjects from the literature are displayed as reported.

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