

Supplementary Table SI. Genotype frequencies and predicted functional effect of selected *CCKAR* and *CCKBR* polymorphisms

SNP	Predicted effect	Genotype	Healthy controls ( <i>N</i> = 82)			PCOS patients ( <i>N</i> = 168)		
			<i>N</i> (%)	MAF	pHWE	<i>N</i> (%)	MAF	pHWE
<i>CCKAR</i> rs6448456 c.-85G>C 5'UTR	Binding of transcription factors and intron splicing	G/G	54 (65.9)	0.189	0.960	114 (67.9)	0.182	0.448
		G/C	25 (30.5)			47 (28.0)		
		C/C	3 (3.7)			7 (4.2)		
<i>CCKAR</i> rs1800857 c.113-5T>C intron	Binding of transcription factors	T/T	58 (70.7)	0.156	0.121	115 (68.5)	0.167	0.355
		T/C	24 (29.3)			50 (29.8)		
		C/C	0 (0.0)			3 (1.8)		
<i>CCKBR</i> rs2929180 c.-51G>T 5'UTR	Binding of transcription factors	G/G	55 (67.1)	0.177	0.669	121 (72.0)	0.170	<b>0.005</b>
		G/T	25 (30.5)			37 (22.0)		
		T/T	2 (2.4)			10 (6.0)		
<i>CCKBR</i> rs1800843 c.559+32C>A intron	Intron splicing	C/C	55 (67.1)	0.171	0.278	108 (65.1) [2]	0.187	0.361
		C/A	26 (31.7)			54 (32.5)		
		A/A	1 (1.2)			4 (2.4)		
<i>CCKBR</i> rs1042047 c.*409A>C 3'UTR	miRNA binding site	A/A	35 (42.7)	0.378	0.123	81 (48.5) [1]	0.347	<b>&lt; 0.001</b>
		A/C	32 (39.0)			56 (33.5)		
		C/C	15 (18.3)			30 (18.0)		
<i>CCKBR</i> rs1042048 c.*437A>G 3'UTR	miRNA binding site	A/A	33 (40.2)	0.341	0.209	80 (48.2) [2]	0.322	0.182
		A/G	42 (51.2)			65 (39.2)		
		G/G	7 (8.5)			21 (12.7)		

SNP – single nucleotide polymorphism, 5'UTR – 5'-untranscribed region, 3'UTR – 3'-untranslated region, MAF – minor allele frequency, p<sub>HWE</sub> – Hardy-Weinberg equilibrium, [] – number of missing data. Statistically significant p-values are shown in bold.

Supplementary Table SII. Association of selected *CCKAR* and *CCKBR* polymorphisms with PCOS susceptibility

Polymorphism	Genotype	Controls <i>N</i> (%)	PCOS <i>N</i> (%)	OR (95% CI)	<i>P</i> -value	OR (95% CI) <sub>adj</sub>	<i>P</i> <sub>adj</sub>
<i>CCKAR</i> rs6448456	G/G	54 (65.9)	114 (67.9)	Reference		Reference	
	G/C+C/C	28 (34.1)	54 (32.1)	0.91 (0.52–1.60)	0.751	1.15 (0.60–2.22)	0.680
<i>CCKAR</i> rs1800857	T/T	58 (70.7)	115 (68.5)	Reference		Reference	
	T/C+C/C	24 (29.3)	53 (31.5)	1.11 (0.63–1.98)	0.714	1.07 (0.54–2.11)	0.843
<i>CCKBR</i> rs2929180	G/G	55 (67.1)	121 (72.0)	Reference		Reference	
	G/T+T/T	27 (32.9)	47 (28.0)	0.79 (0.45–1.40)	0.421	0.82 (0.42–1.62)	0.574
<i>CCKBR</i> rs1800843	C/C	55 (67.1)	108 (65.1)	Reference		Reference	
	C/A+A/A	27 (32.9)	58 (34.9)	1.09 (0.63–1.92)	0.753	1.42 (0.74–2.75)	0.295
<i>CCKBR</i> rs1042048	A/A	33 (40.2)	80 (48.2)	Reference		Reference	
	A/G+G/G	49 (59.8)	86 (51.82)	0.72 (0.42–1.24)	0.238	0.93 (0.50–1.75)	0.825

PCOS – polycystic ovary syndrome, OR – odds ratio, CI – confidence interval, adj – adjusted for age.

Supplementary Table SIII. *CCKAR* polymorphisms and anthropometric characteristics of patients with polycystic ovary syndrome ( $N = 168$ )

Characteristics	Genotype*	<i>CCKAR</i> rs6448456		<i>CCKAR</i> rs1800857	
		Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value
Body weight [kg] [5]	X/X	100.9 (86.9–112.3)	0.245	99.7 (85–110.8)	0.410
	X/x+x/x	97.3 (82.6–107.5)		101 (84.3–111.4)	
BMI [kg/m <sup>2</sup> ] [6]	X/X	36.2 (32–40.6)	0.143	35.8 (31.3–39.7)	0.362
	X/x+x/x	34.3 (30.9–38.5)		35.5 (32–41.1)	
Waist circumference [cm] [31]	X/X	112 (102–123)	0.591	110 (100.5–121.3)	0.303
	X/x+x/x	111 (98.8–121)		113.5 (102.3–122.3)	
VAT area [cm <sup>2</sup> ] [74]	X/X	159 (119–205)	0.285	146.5 (108.8–197.8)	0.244
	X/x+x/x	145 (113–179.5)		167.5 (127–201)	

\*X – major (common) allele, x – minor (variant) allele, BMI – body mass index, VAT – visceral adipose tissue, [] – number of missing data.

Supplementary Table SIV. *CCKBR* polymorphisms and anthropometric characteristics of patients with polycystic ovary syndrome ( $N = 168$ )

Characteristics	Genotype*	<i>CCKBR</i> rs2929180		<i>CCKBR</i> rs1800843		<i>CCKBR</i> rs1042048	
		Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value
Body weight [kg] [5]	X/X	100 (85–111.6)	0.933	101 (84.5–111.7)	0.503	100 (86.6–110.1)	0.985
	X/x+x/x	101 (84.9–110.9)		96.2 (85.2–111.1)		100.9 (85–114)	
BMI [kg/m <sup>2</sup> ] [6]	X/X	35.7 (31.9–40.3)	0.704	35.9 (31.5–39.9)	0.836	36.3 (32.4–39.9)	0.781
	X/x+x/x	36.6 (31.5–39.1)		35.8 (31.8–40.8)		35.6 (30.9–40.3)	
Waist circumference [cm] [31]	X/X	110.3 (101.8–123)	0.917	110 (101–122)	0.412	112 (105.1–122.9)	0.371
	X/x+x/x	114 (102–121)		113.5 (103.8–121)		109.5 (100–121)	
VAT area [cm <sup>2</sup> ] [74]	X/X	148 (115.5–203)	0.402	150 (116–186.5)	0.510	165 (135–194)	0.329
	X/x+x/x	165 (132.5–189)		157.5 (118.5–208)		147.5 (114–201)	

\* X – major (common) allele, x – minor (variant) allele. BMI – body mass index; VAT – visceral adipose tissue; [] – number of missing data.

Supplementary Table SV. *CCKAR* polymorphisms and anthropometric characteristics of healthy controls ( $N = 82$ )

Characteristics	Genotype*	<i>CCKAR</i> rs6448456		<i>CCKAR</i> rs1800857	
		Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value
Body weight [kg]	X/X	61.4 (55.2–70.8)	0.214	62.2 (55.2–72.2)	0.819
	X/x+x/x	64.1 (57.7–72.3)		61.6 (56.7–73)	
BMI [kg/m <sup>2</sup> ]	X/X	22.1 (19.8–24.6)	0.205	22.5 (20.2–25)	0.695
	X/x+x/x	22.7 (20.8–25.4)		22.2 (20.1–25)	
Waist circumference [cm]	X/X	73 (66–77.3)	0.419	73 (66.8–79)	0.874
	X/x+x/x	73.5 (67.5–80.5)		73 (66.3–78)	
Visceral fat (arbitrary units)	X/X	3 (2–4)	0.217	3.5 (3–4.3)	0.314
	X/x+x/x	3.5 (3–5)		3 (2.3–4)	
Fat mass (%)	X/X	30.4 (25.8–35.9)	0.172	32.6 (26.8–37.1)	0.650
	X/x+x/x	33.3 (27.6–37.9)		30.4 (26.1–37.1)	
Fat free mass (%)	X/X	28.8 (27.5–30.8)	0.148	28.3 (27.1–30)	0.551
	X/x+x/x	28.1 (25.9–29.5)		28.8 (26.6–30.9)	

\*X – major (common) allele, x – minor (variant) allele, BMI – body mass index.

Supplementary Table SVI. *CCKBR* polymorphisms and anthropometric characteristics of healthy controls (*N* = 82)

Characteristics	Genotype*	<i>CCKBR</i> rs2929180		<i>CCKBR</i> rs1800843		<i>CCKBR</i> rs1042048	
		Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value	Median (25–75%)	<i>P</i> -value
Body weight [kg]	X/X	62.1 (55–70.4)	0.329	60.8 (55–70.3)	0.077	62.1 (54.3–74.5)	0.925
	X/x+x/x	63.6 (57–76.7)		65.7 (58–77.1)		62.2 (56.7–71.3)	
BMI [kg/m <sup>2</sup> ]	X/X	22.1 (19.6–24.8)	0.135	22 (19.6–24.9)	0.071	22.3 (19.4–25.5)	0.828
	X/x+x/x	22.4 (21.1–27.1)		23.5 (21.6–25.2)		22.4 (20.8–24.9)	
Waist circumference [cm]	X/X	71 (66–78)	0.222	69 (66–78)	<b>0.027</b>	73 (66.5–80)	0.809
	X/x+x/x	73 (69–82)		74 (71–78)		73 (66.5–78)	
Visceral fat (arbitrary units)	X/X	3 (2–4)	0.143	3 (2–4)	<b>0.046</b>	4 (2.5–5)	0.502
	X/x+x/x	3 (3–5)		4 (3–5)		3 (3–4)	
Fat mass (%)	X/X	31.6 (26.3–35.6)	0.319	30.2 (25.5–37.1)	0.109	32.7 (25.5–37.1)	0.755
	X/x+x/x	32.8 (26.3–38.2)		33.9 (30.5–36.6)		32.2 (26.7–36.9)	
Fat free mass (%)	X/X	28.5 (27.5–30.4)	0.453	28.9 (27.1–30.8)	0.364	28.3 (27.2–30.8)	0.992
	X/x+x/x	28.3 (25.5–31.5)		28.3 (26.7–29.3)		28.7 (26.7–30.3)	

\*X – major (common) allele, x – minor (variant) allele, BMI – body mass index. Statistically significant p-values are printed in bold.