

Erasmus MC

Universitair Medisch Centrum Rotterdam



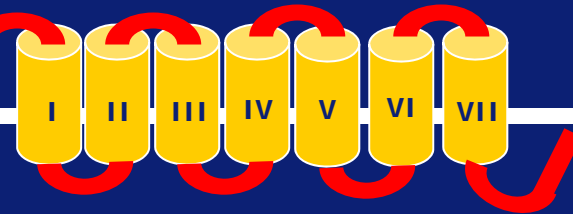
Genetics of PCOS: gonadotrophic hormone receptors

O. Valkenburg

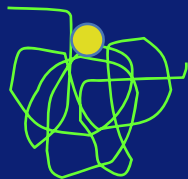
**Y. Louwers, Sharon Lie Fong, Jits Schipper, Evert JP van Santbrink, Marinus JC Eijkemans,
Bart CJM Fauser, Axel NP Themmen, Frank H de Jong, André G Uitterlinden, Aida M Bertoli-
Avella, Ben A Oostra en Joop SE Laven**

FSH receptor polymorphism

680 Asn/Ser (exon 10)



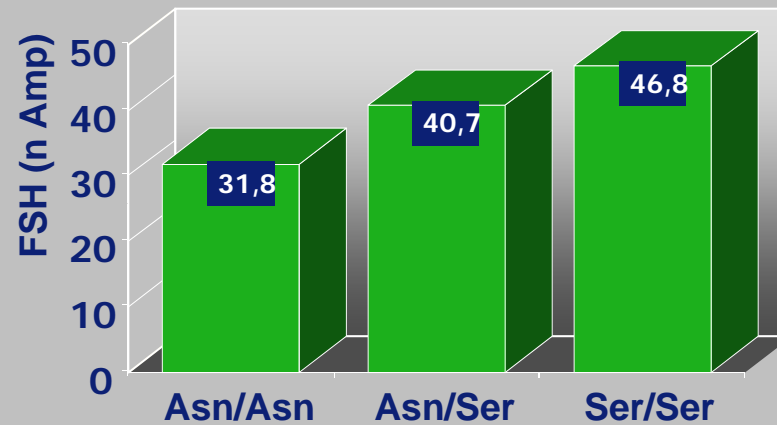
..ACGAATA



Asn

ATA..

Ser

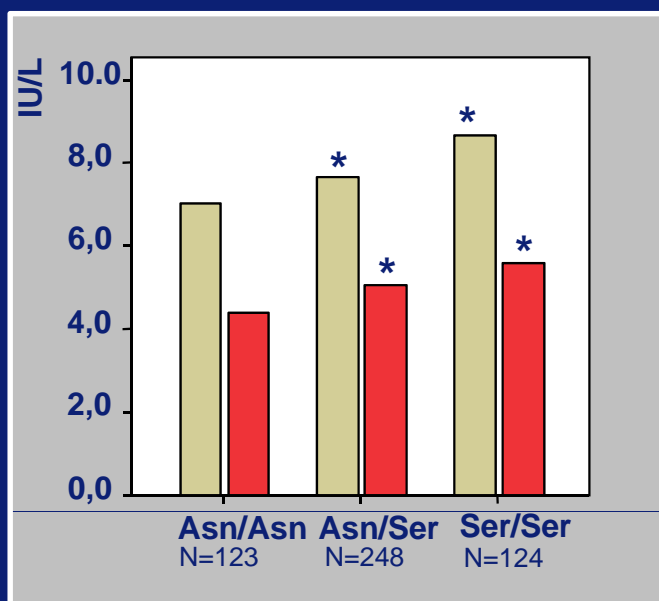


Perez Mayorga M et al., JCEM, 2000

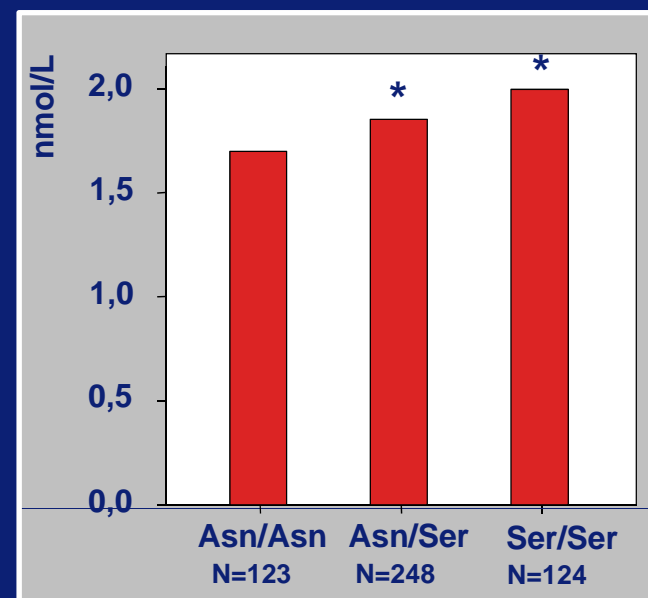
PCOS phenotype

FSHR polymorphism affects the phenotype

Gonadotrophic hormone levels



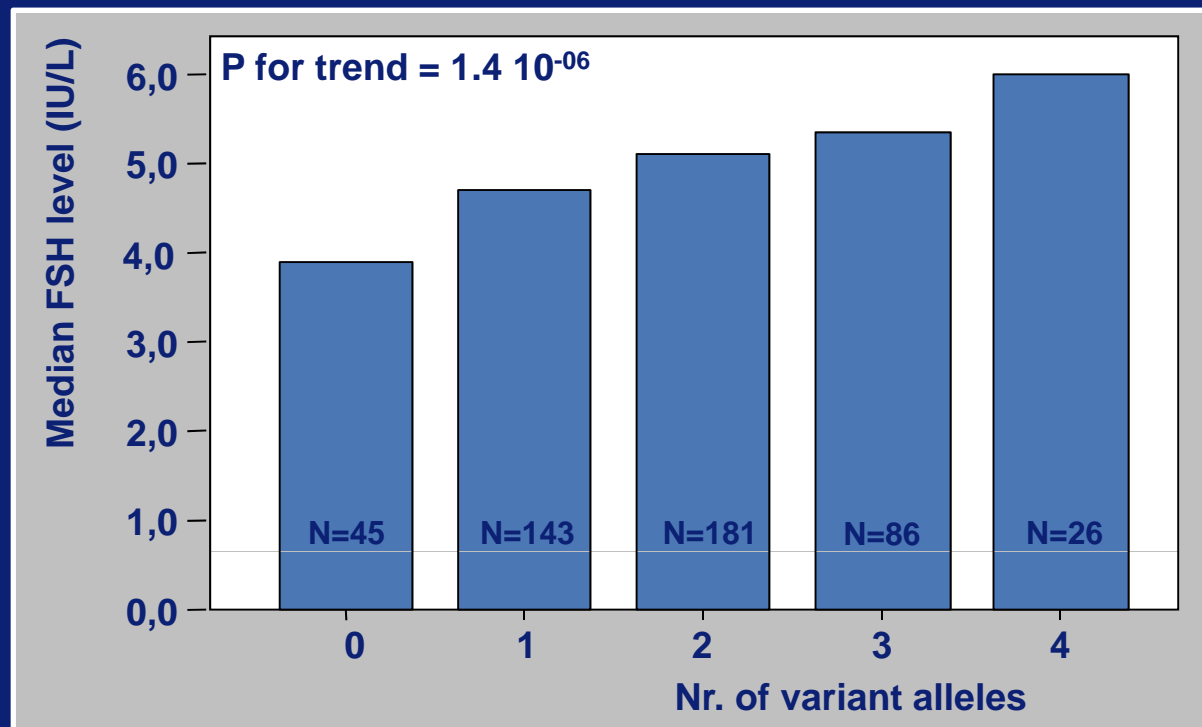
Testosterone levels



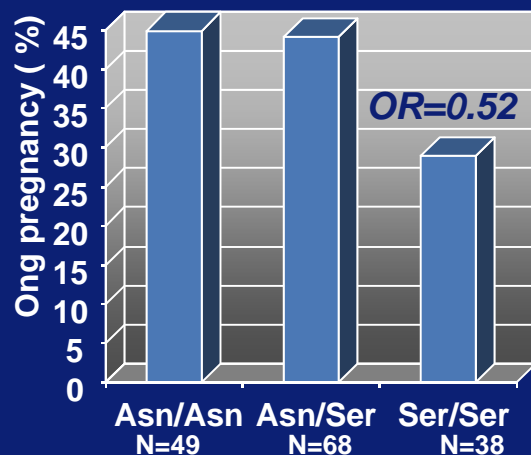
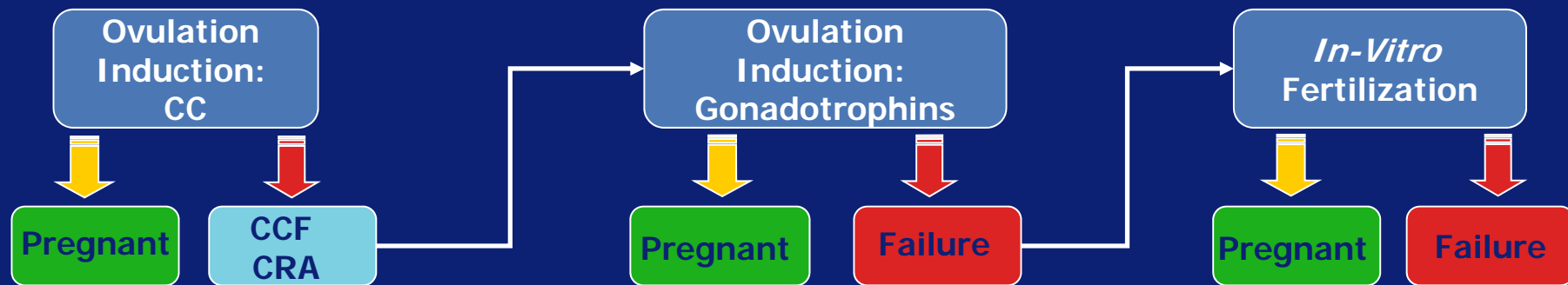
PCOS Phenotype

Combined FSHR 680 Asn/Ser and LHR 312 Ser/Asn polymorphisms

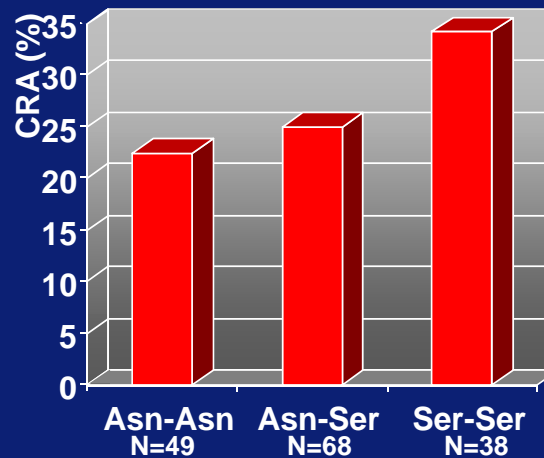
Risk allele score: basal FSH levels



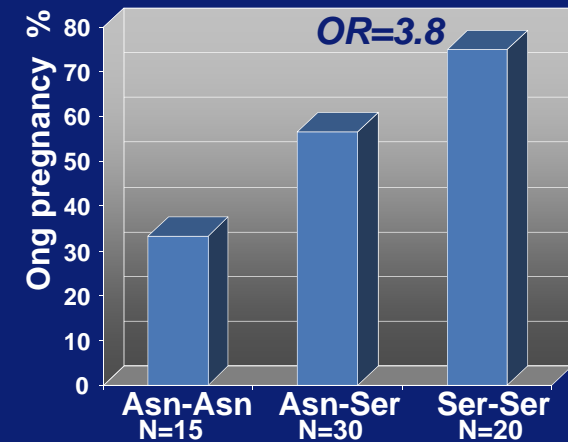
Treatment: WHOII anovulation



Ong pregnancy (CC)



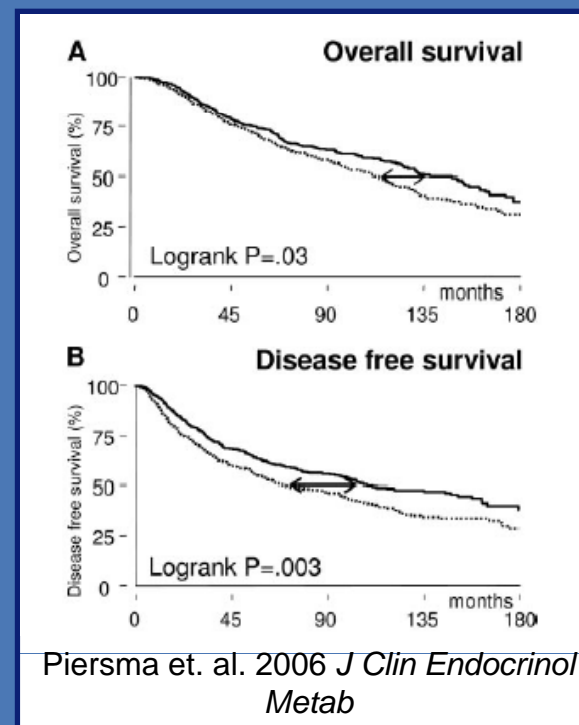
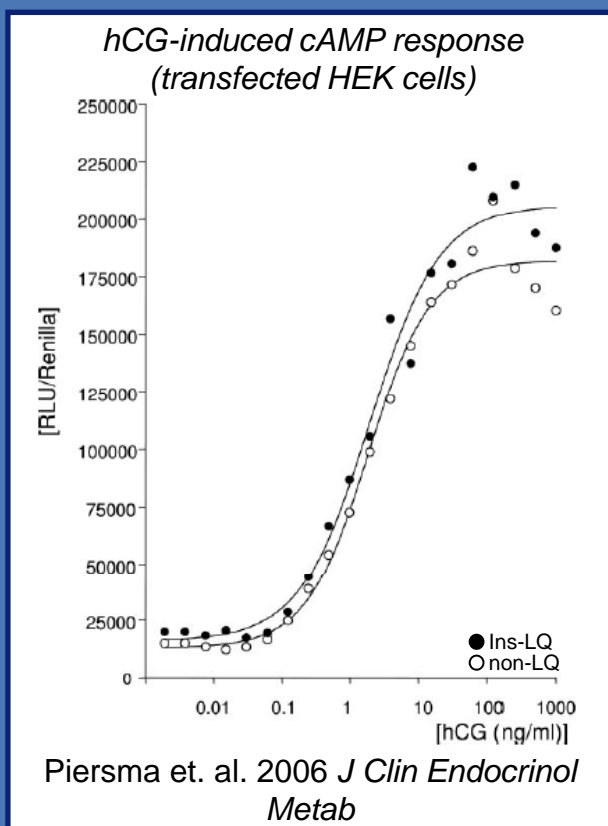
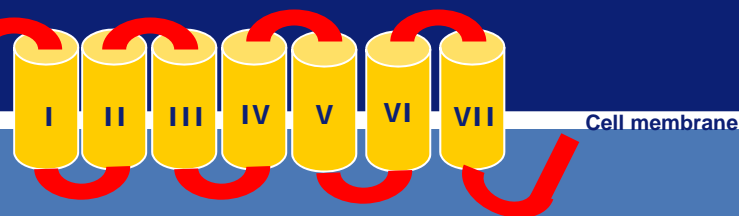
Clomiphene res. anovulation



Ong pregnancy (FSH)

LH receptor polymorphism

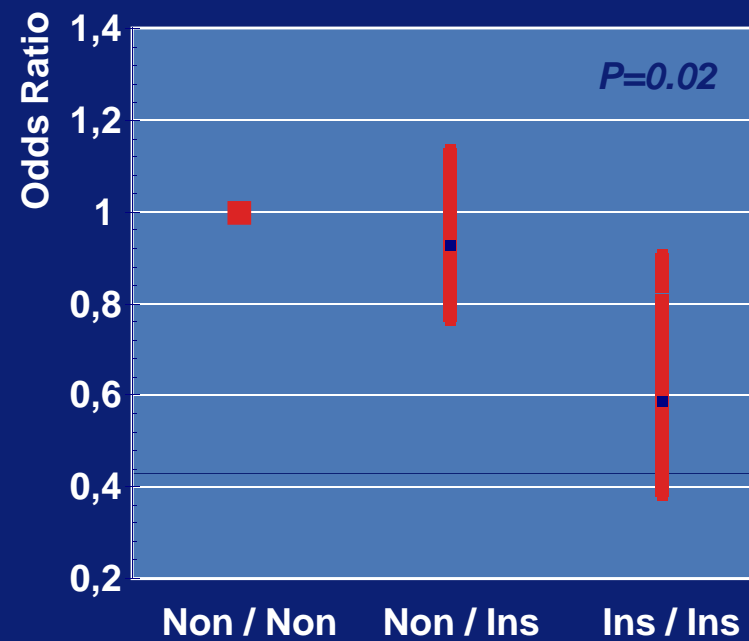
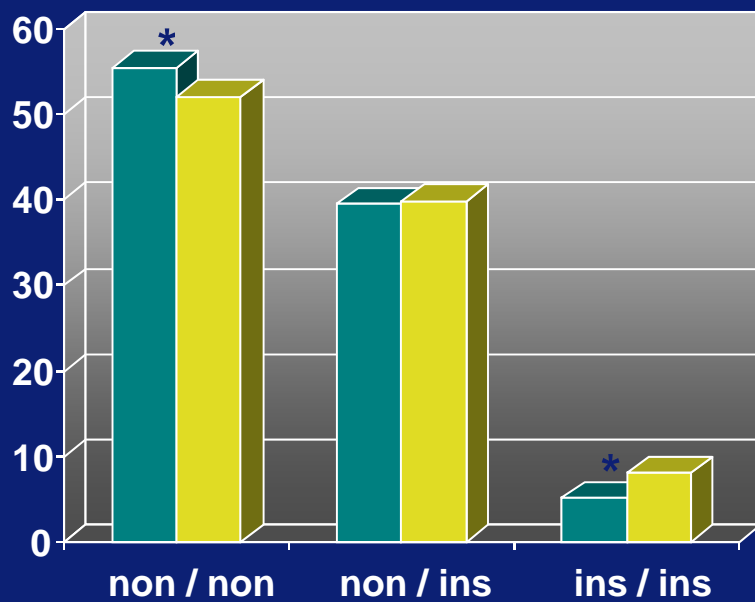
LH-R insLQ



PCOS phenotype

LH-R insLQ polymorphism affects PCOS risk

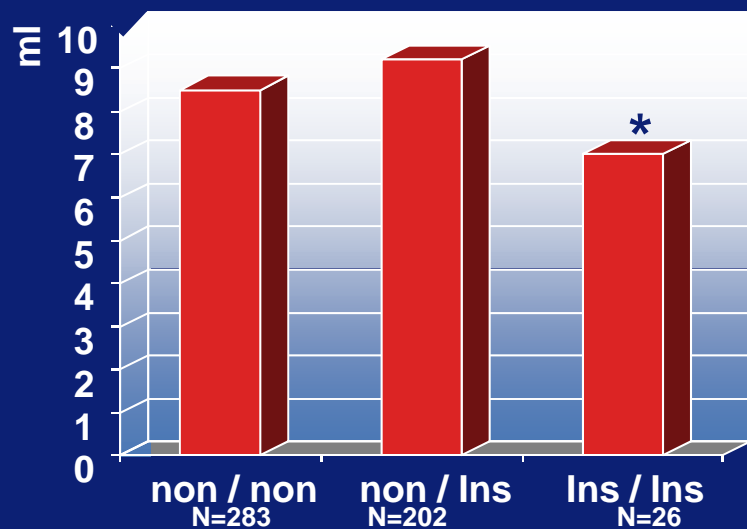
■ PCOS (n=518) ■ Control (n=2996)



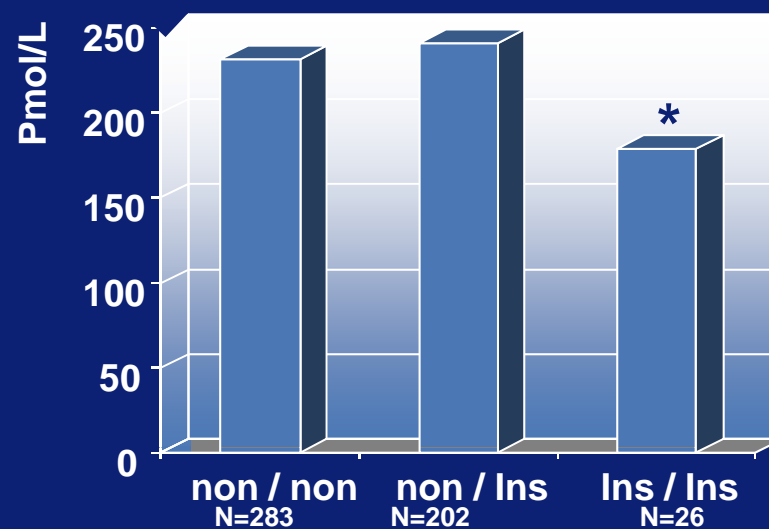
LH receptor polymorphism

LH-receptor insLQ polymorphism affects the phenotype of PCOS

Mean ovarian volume



Estradiol levels



Conclusions

FSH receptor 680 Asn / Ser polymorphism:

- is associated with a higher setpoint for gonadotrophic hormones (FSH and LH).
- can be used as a predictor of outcome of ovulation induction (clomiphene citrate and FSH).

LH receptor ins-LQ polymorphism:

- possibly protects for PCOS in homozygous carriers (needs confirmation)
- is associated with lower ovarian volume and estradiol levels in homozygous carriers.

Thanks to...

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