

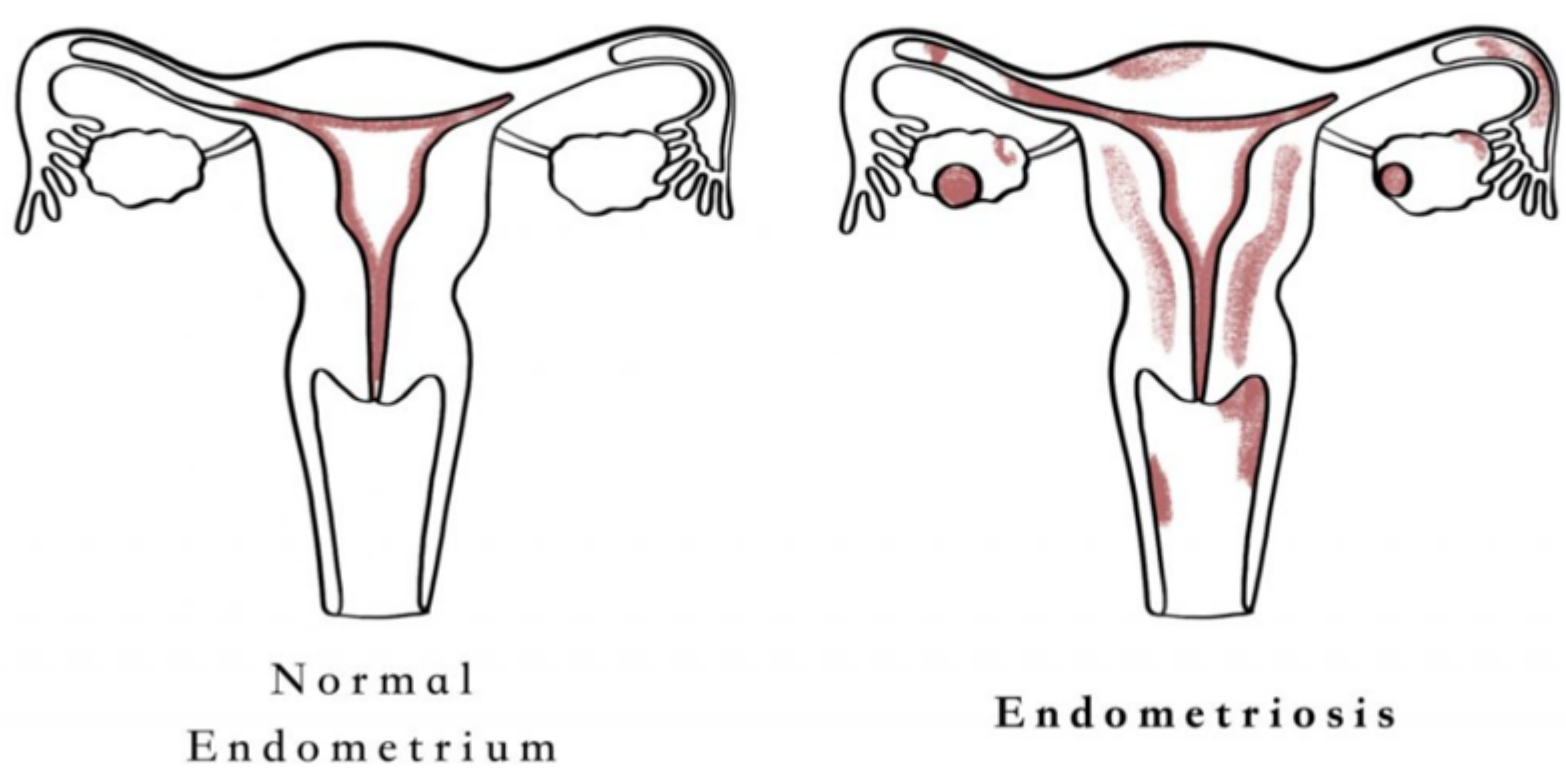
The pathogenesis of endometriosis

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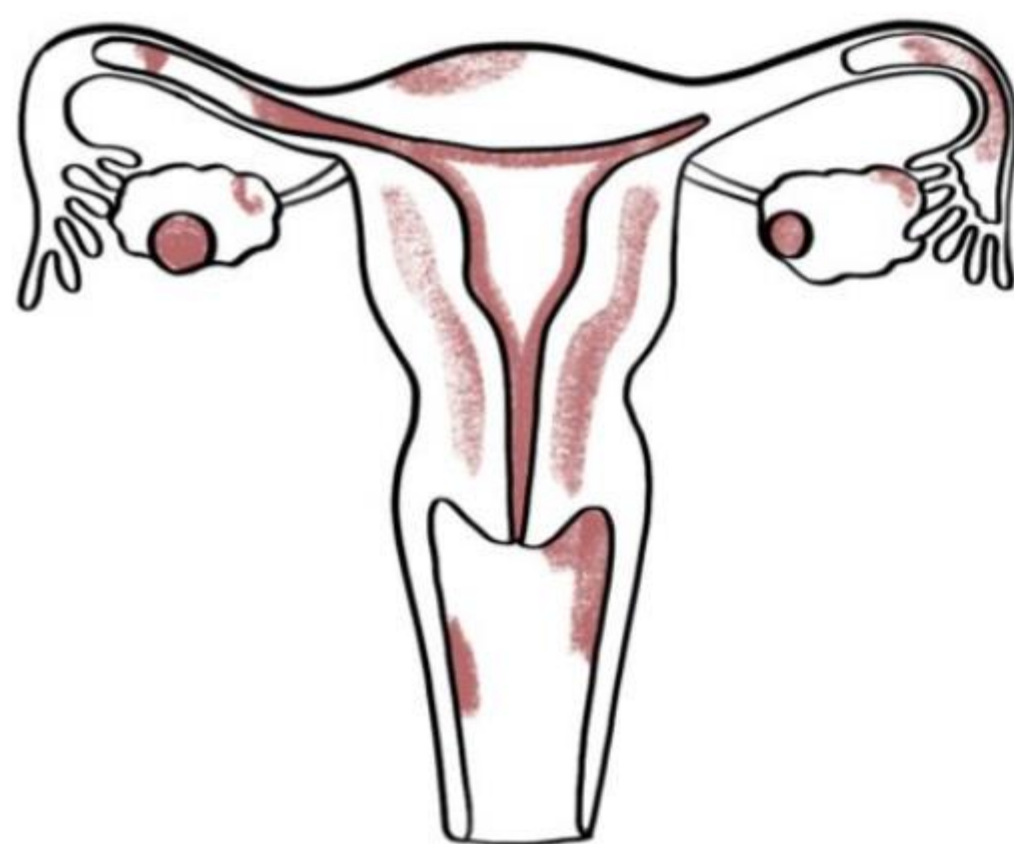
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Endometriosis

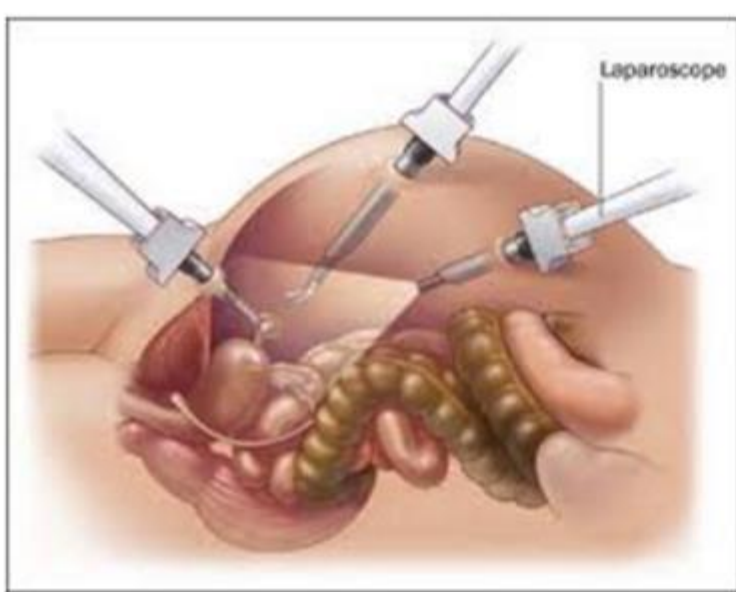
- Definition : the growth of endometrial tissue outside the uterine cavity
- Chronic estrogen-dependent inflammatory disease
- Associated with dysmenorrhea, pelvic pain and infertility
 - 6% to 10% of women with endometriosis (~200 million worldwide)
 - ~50-70% of women with pelvic pain
 - ~15-50% of women with infertility
- Surgical therapy targets visible lesion
 - recurrence rate of > 50% after surgical resection (within 2 years)



Challenge of endometriosis



■ Diagnosis: Laparoscopic surgery

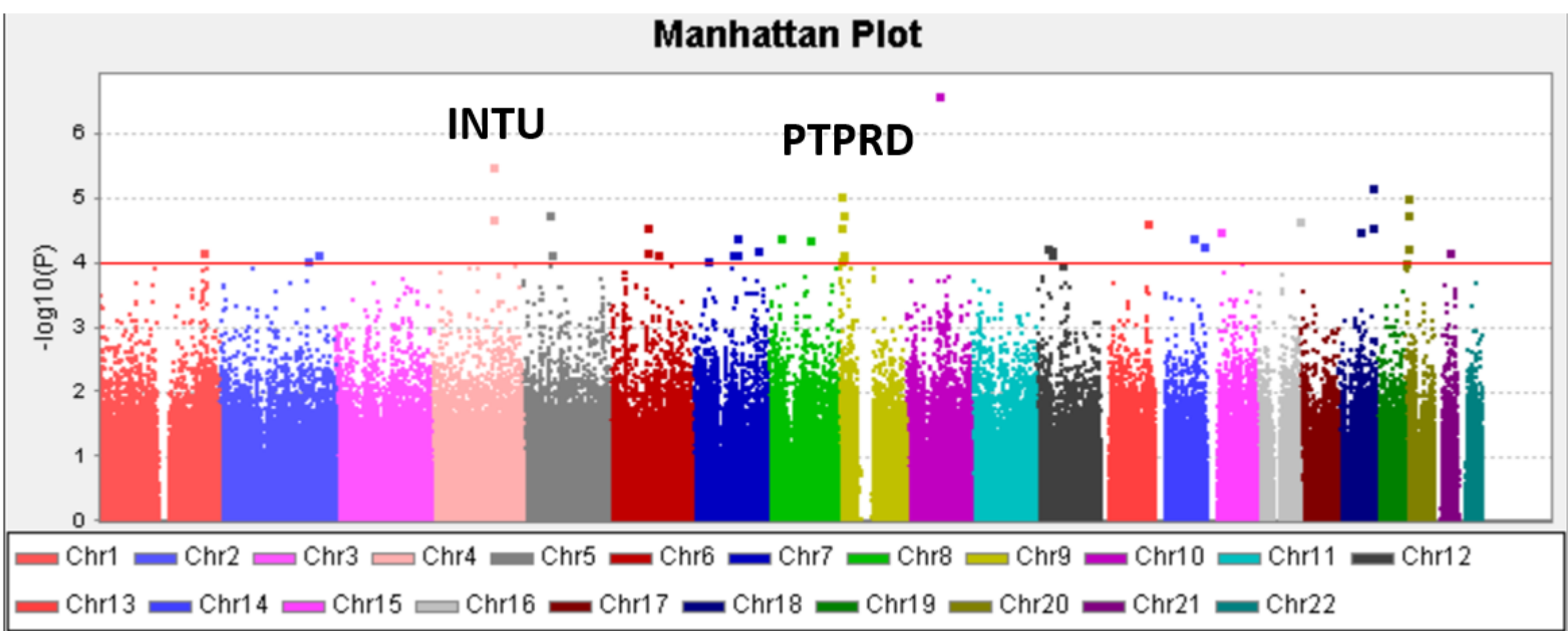


- There is no non-invasive diagnosis tests
- There are no disease-specific medicine
- Treatment failure is common

Lancet (2004)364:1789
Cell (2021)184:2807

Genome-wide association study (GWAS) of endometriosis

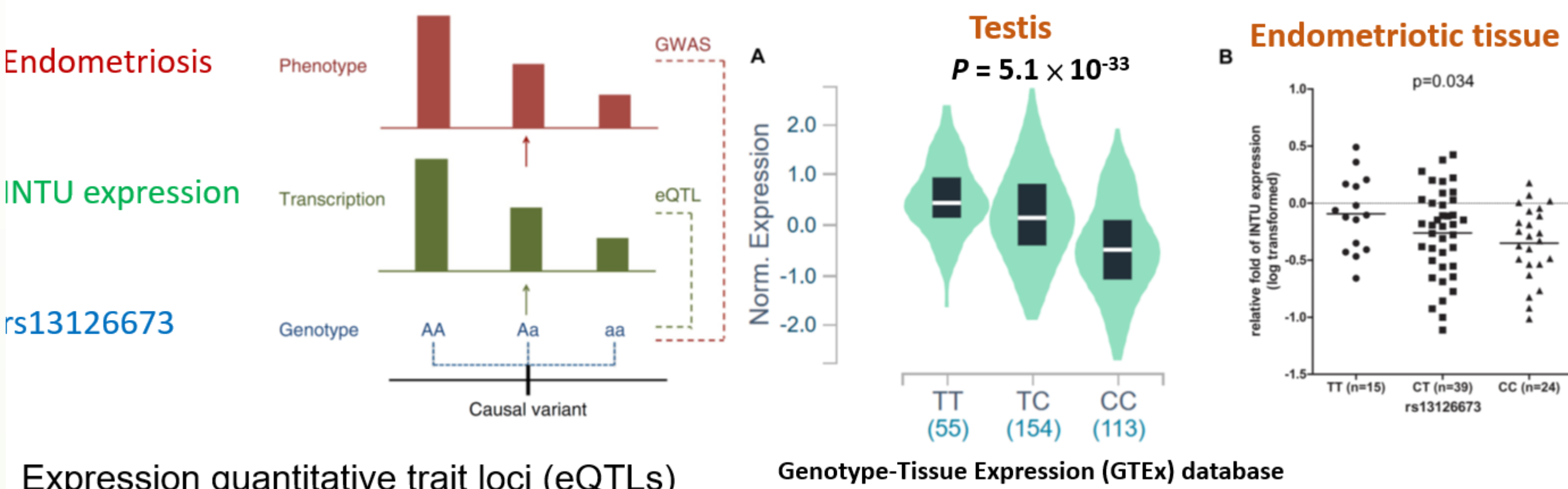
Laproscopy and histological confirmed Extracted blood DNA	GWAS	Cases (n)	Controls (n)	Cochran-Armitage trend test Case/ Control association study
	Taiwan biobank (TWB) chip 650,000 SNP	126	96	
	Replication	133	75	
	Combine	259	171	
Total : 430				



INTU: intuned planar cell polarity protein
PTPRD: protein tyrosine phosphatase, receptor type, D

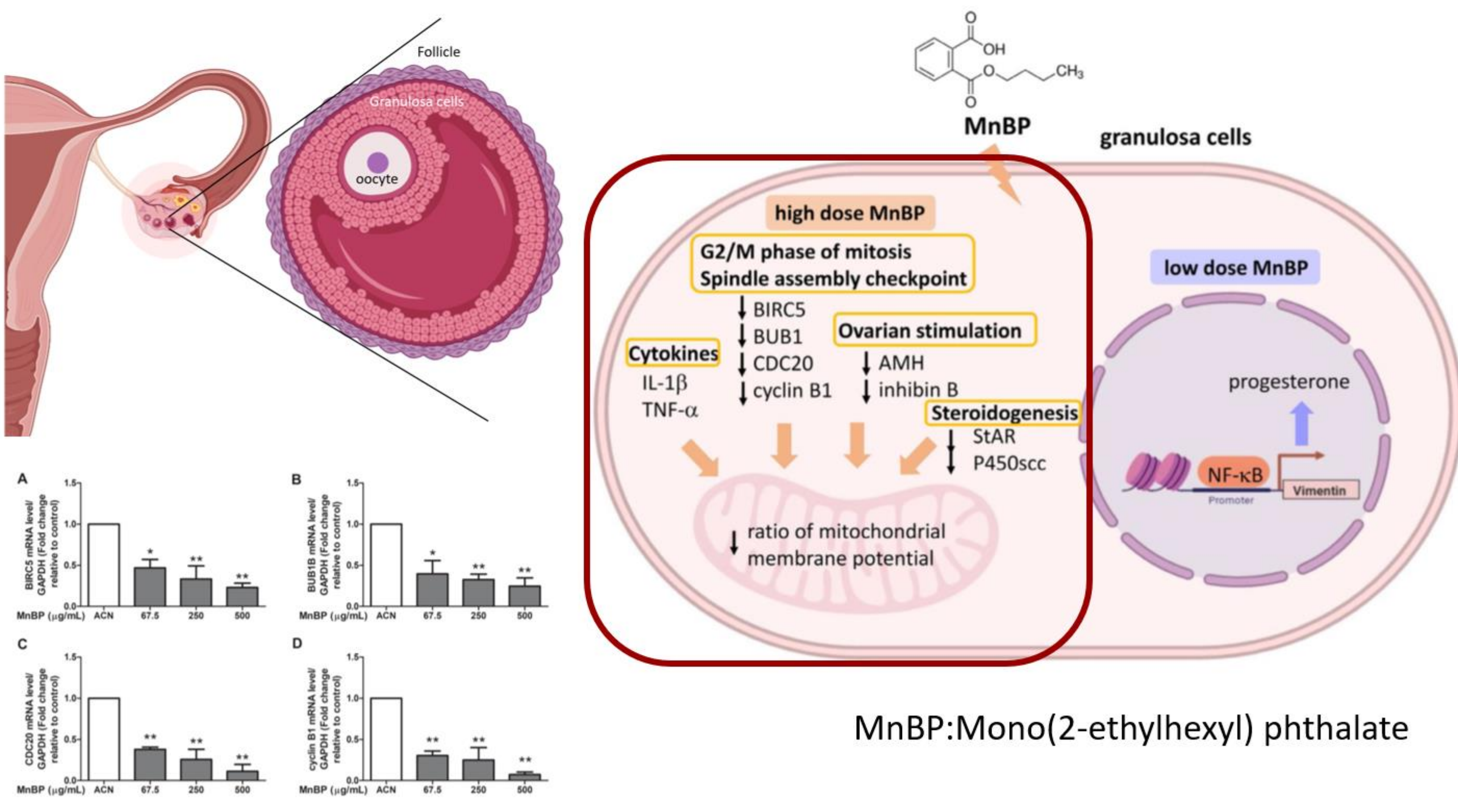
Integration of genome-wide association study (GWAS) and expression quantitative trait loci (eQTL)

Chr.	Position	SNP	Gene	Allele 1	Allele 2	Risk Allele	RAF controls	RAF cases	Discovery Trend P	Risk allele OR	95% CI
4	128586241	rs13126673	INTU	T	C	C	0.4793	0.6142	1.10E-04	1.729	1.309 2.284
5	52363759	rs2056401	ITGA2	G	A	A	0.7735	0.8643	4.76E-04	1.865	1.304 2.668
7	90650256	rs7789771	CDK14	G	A	G	0.04734	0.1152	7.21E-04	2.621	1.481 4.638
7	90660684	rs1358101	CDK14	A	C	A	0.04706	0.1148	7.03E-04	2.626	1.484 4.646
9	9707144	rs10739199	PTPRD	G	A	A	0.6265	0.7578	6.75E-05	1.866	1.384 2.515
9	9733309	rs2025392	PTPRD	T	C	C	0.8824	0.9553	8.01E-05	2.846	1.671 4.848
9	9734220	rs2761699	PTPRD	C	T	T	0.8853	0.9512	4.05E-04	2.524	1.497 4.255



Expression quantitative trait loci (eQTLs)
Nature Genetics (2016)48:481

The effects of MnBP on human granulosa cells



The treatment of gonadotropin-releasing hormone agonist (GnRHa) in women with endometriosis

