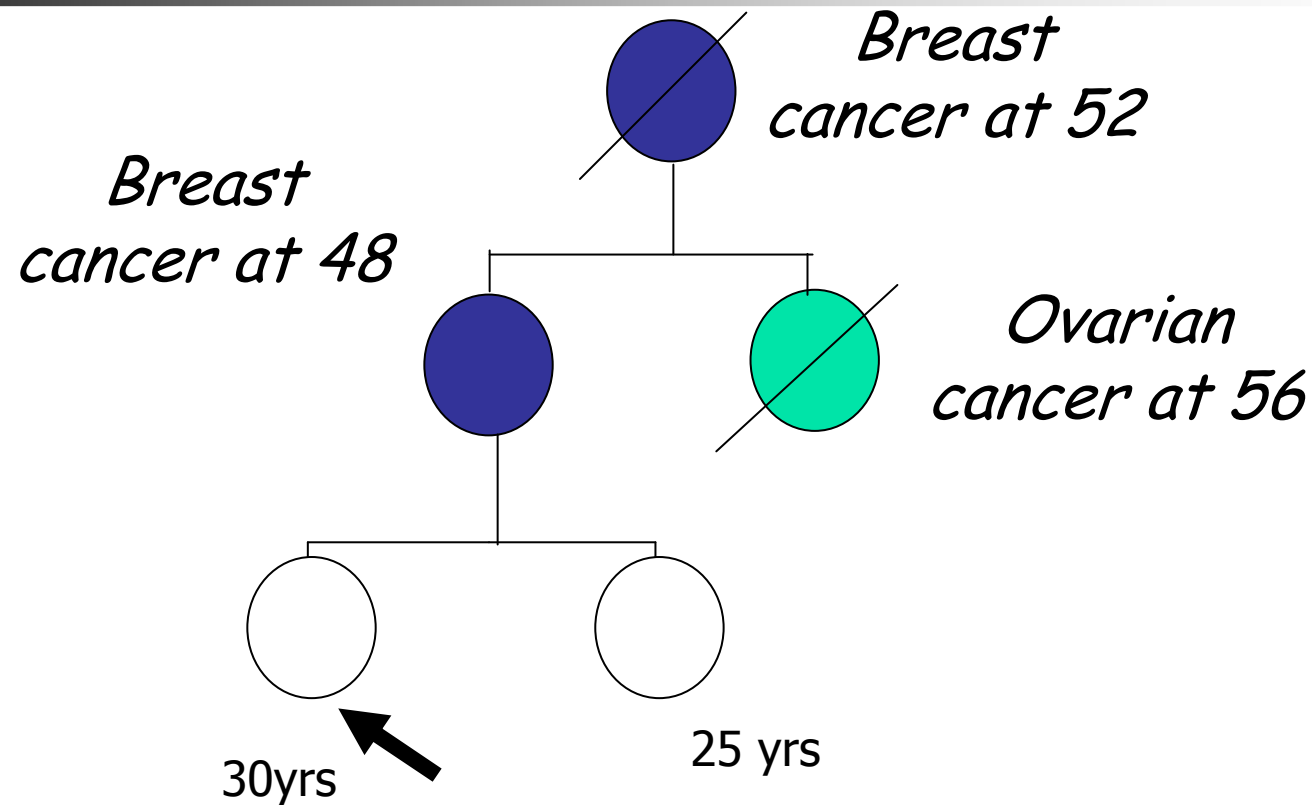


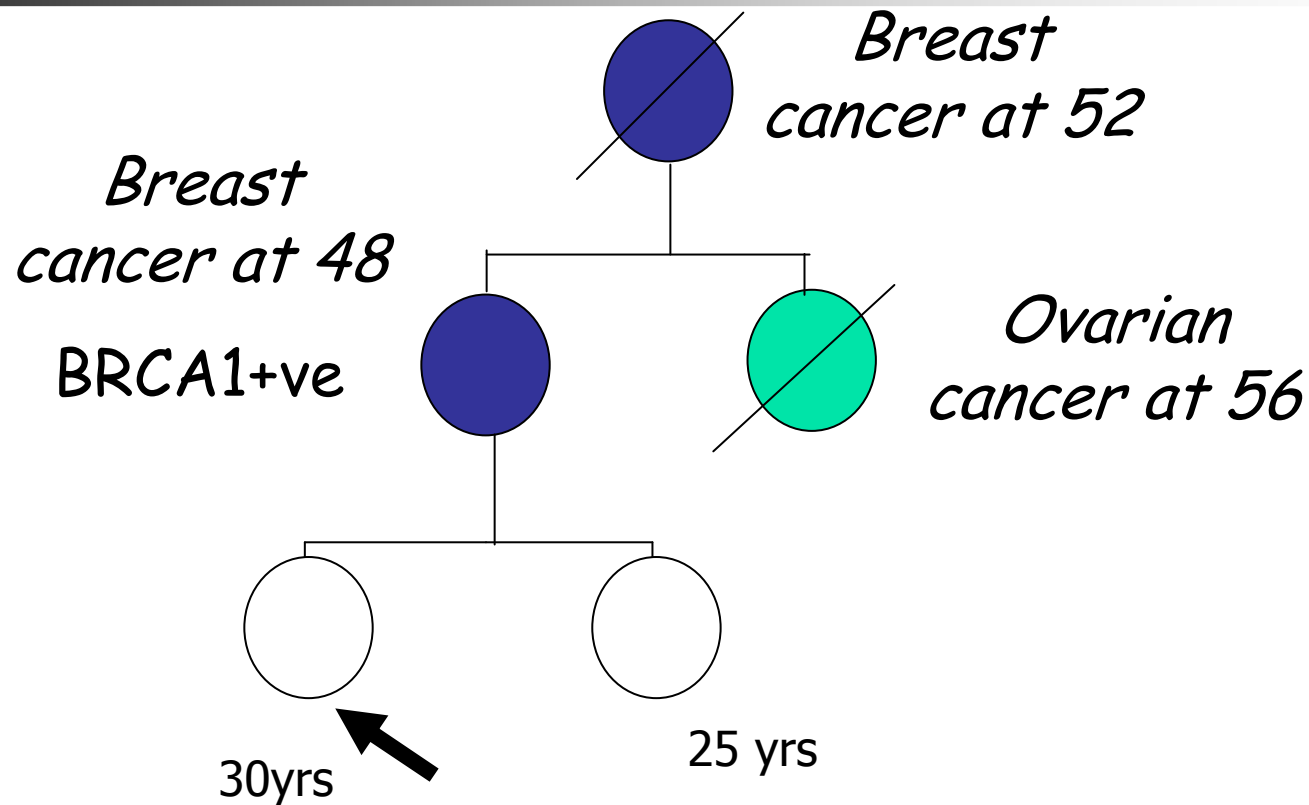
Cancer genetics

Dr Imran Rafi
GPWSI Genetics
Senior Lecturer Primary Care Education
St George's
University of London

Skills Knowledge and attitudes



Skills Knowledge and attitudes



Genes and phenotype

Genes associated with cancer

- BRCA1, BRCA2 → ■ Breast/Ovarian
- APC, MYH → ■ Familial adenomatous polyposis
- MLH1, MSH2, MSH6 → ■ HNPCC
- RET → ■ Medullary thyroid cancer
- P53 → ■ Multiple tumours
- PTEN → ■ Cancer syndromes



Organisation of services

- 'Delivering cancer genetic services – new ways of working*'
 - 7 pilot Macmillan/DH funded projects
 - Kenilworth model
 - Based on Calman-Hine model
 - Common themes for provision of cancer genetics services
 - * Eeles R et al(2007) Familial Cancer 6:163-167(2007)

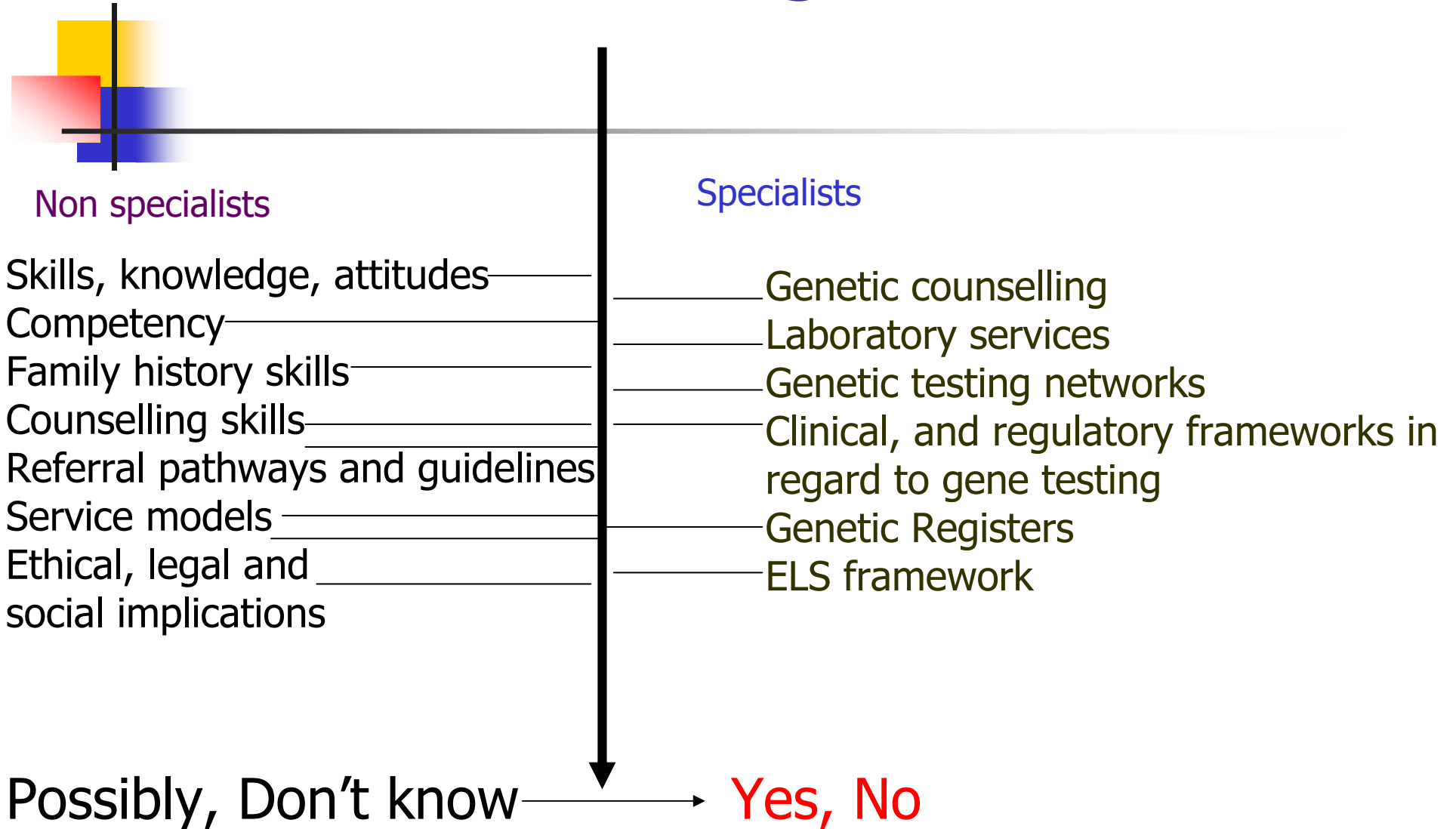


Organisation of services

GPWSI model

- Consensus agreed with regional genetics centre on how referrals are triaged
- Local clinic and infrastructure set up in cottage hospital with local PCT support
- Guidelines for GPs
- Over 80% patients discharged
- Patient and GP satisfaction

Can I have a genetic test?



Genes and Phenotype Management

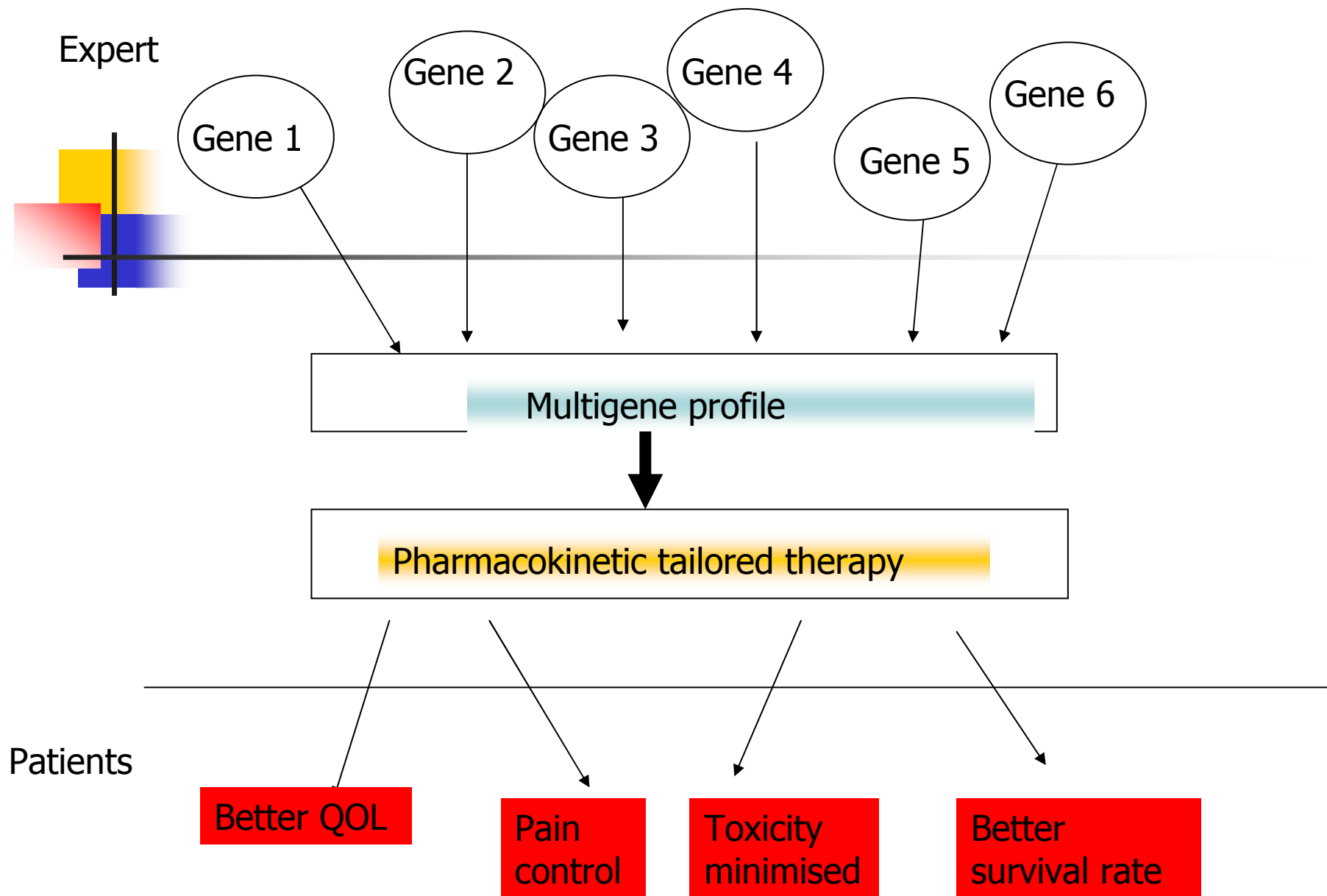


- 'You are a carrier for an altered **APC** gene which you have inherited. You would benefit from colectomy
- 'This women with an altered **BRCA1** gene is requesting bilateral mastectomy to prevent her getting breast cancer'



Pharmacogenetics

- Past
 - This lady has breast cancer
 - She would benefit from surgery and radiotherapy
- Now
 - 'This lady's breast cancer shows ER+, PG - and HER2/neu+ status.
 - She would benefit from Herceptin
- Future
 - 'Full' tumour and patient gene profile
 - She will benefit from this combination of drugs.



Adapted from Davies S et al Haematology 2006

'Clearly relating genotype to phenotype will be one of the great challenges for genetic medicine over the next century'



Professor Sir David Weatherall

- Biobanks
- Wellcome Trust Case Control Consortium

Ethical legal and social

'Over the counter testing'

- 'I can get a genetic test for £500 over the web, should I?'
- 'I would like a genetic test after seeing that TV programme'

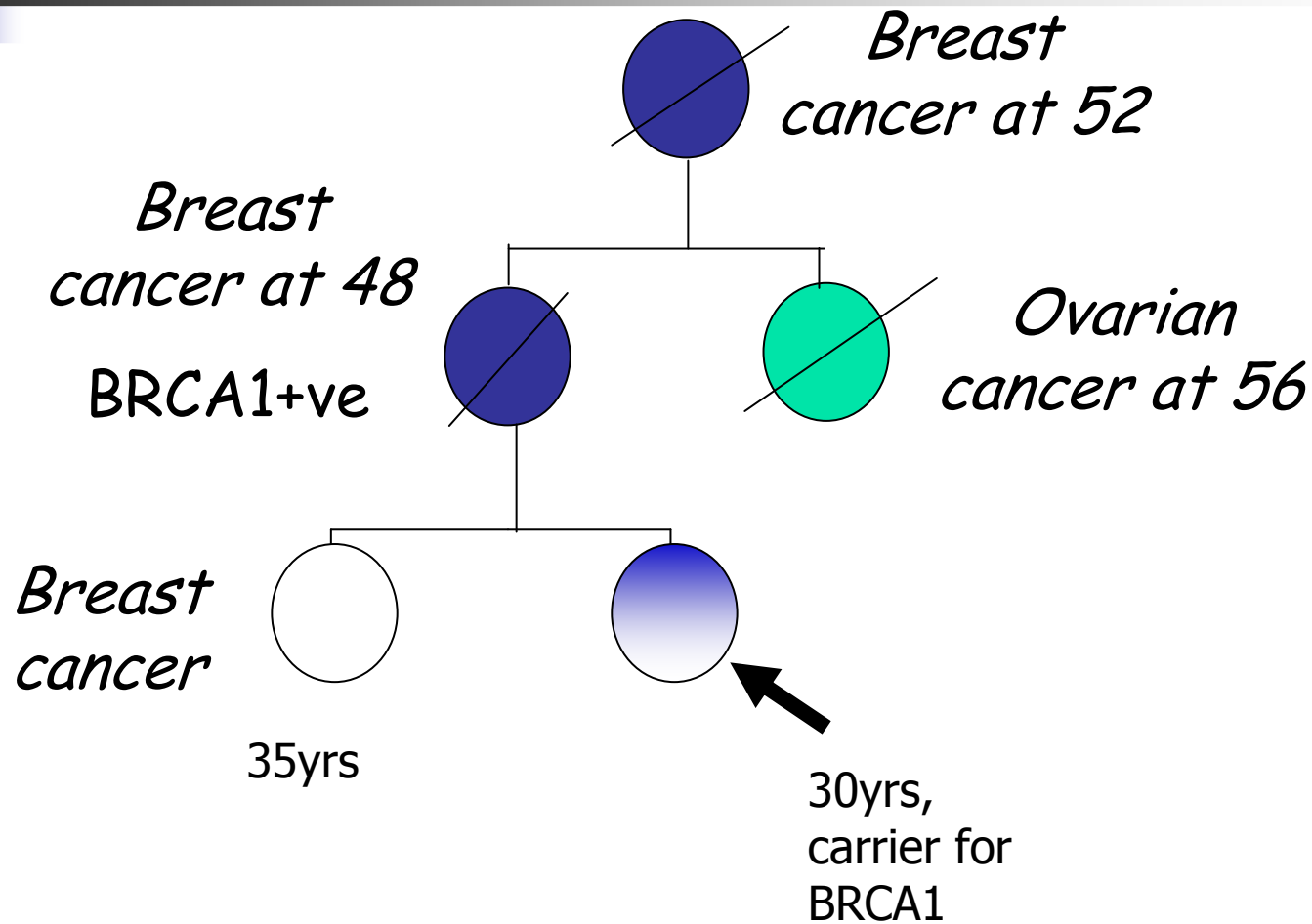




Screening/early detection

- UK Familial Ovarian Cancer Screening Study (UKFOCSS).
 - Is ovarian screening beneficial for women with strong family histories
 - Is ovarian screening beneficial in women with altered BRCA1 or BRCA2 genes?
- Prostate cancer
 - Urinary PCA3

Preimplanation genetic diagnosis



Advances

Clinical genetics

Clinical

- What is known
- What could be improved

Challenges

- Ethics
- Legal
- Social



Conclusion



- Genetic awareness
- Education
- Integration
- Manage demand