

Germline Predisposition to Haematological Malignancies National Consensus Meeting Day 2, April 29th 2022

Dr Terri McVeigh

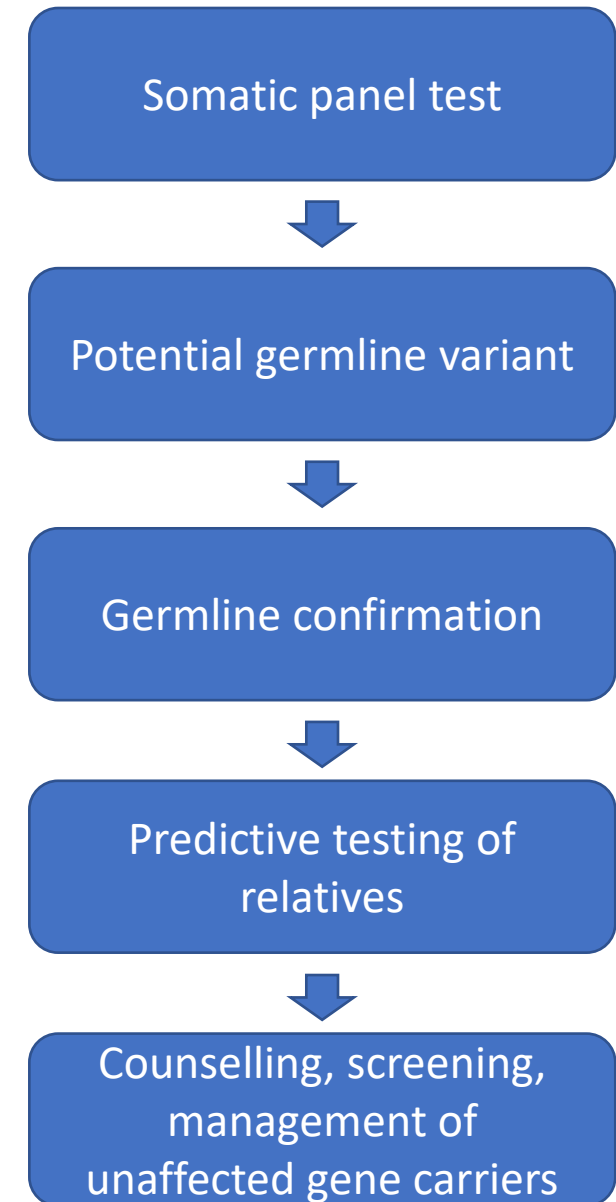
Consultant Clinical Geneticist

Royal Marsden NHS Foundation Trust

Communications Rep, UK Cancer Genetics Group

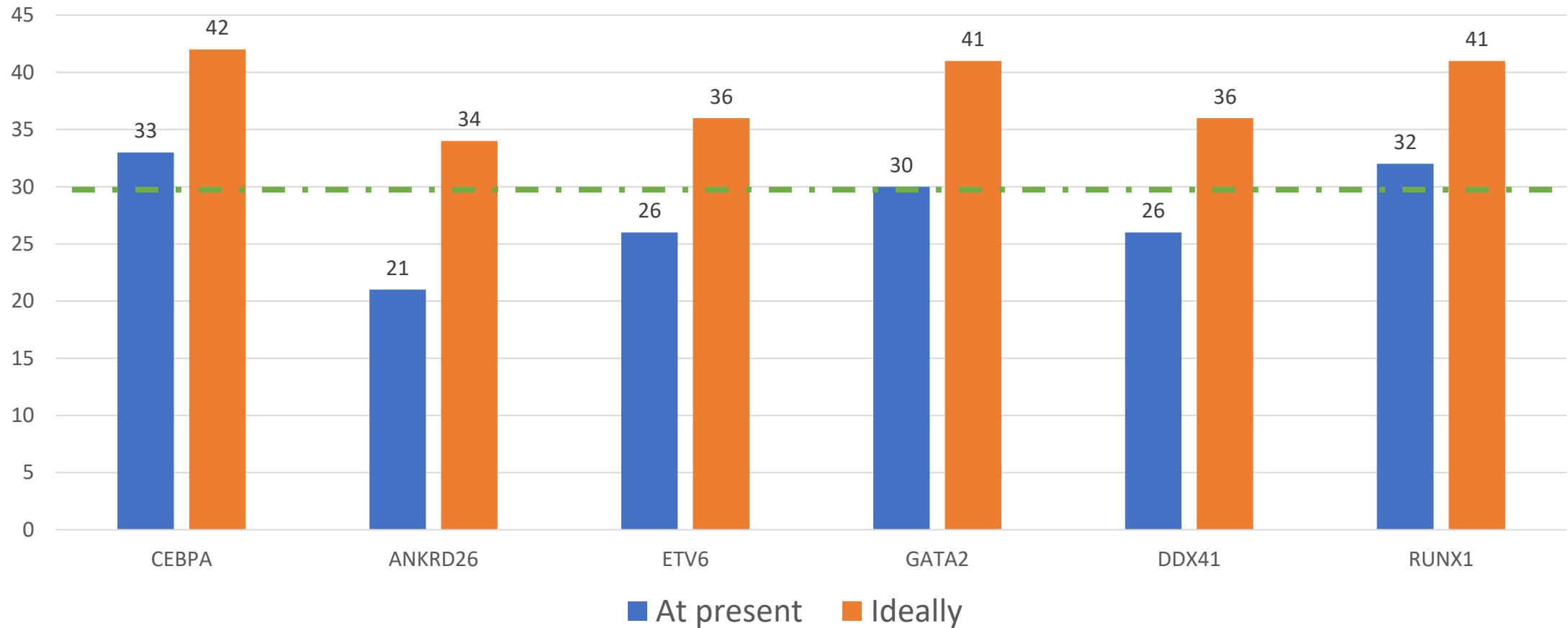
Recap: remit of meeting

- **Focussed** session
 - Guidance on what/how/when we should undertake germline confirmation of somatically identified variants
 - Gene-specific standardised guidelines for germline variant interpretation in genes predisposing to haematological malignancies
 - The requirement for an infrastructure to collect genotype : phenotype data through a national register
 - Clear gene-specific guidelines on clinical management and iterative review of guidelines as on-going data collection informs genotype/phenotype information



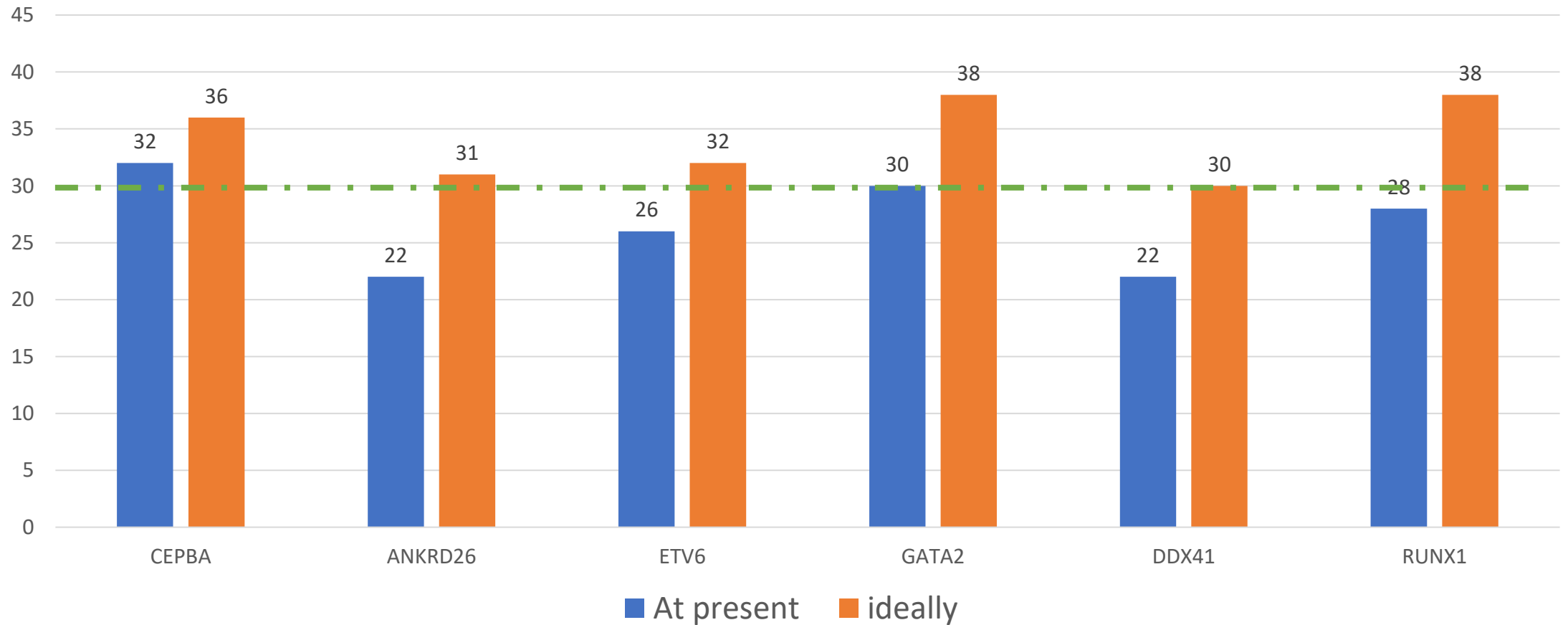
Pre-meeting Survey results (part 2)

Confirmatory germline testing is offered where a likely pathogenic/pathogenic variant of potential germline origin is identified on somatic (tumour-only) testing in the following genes



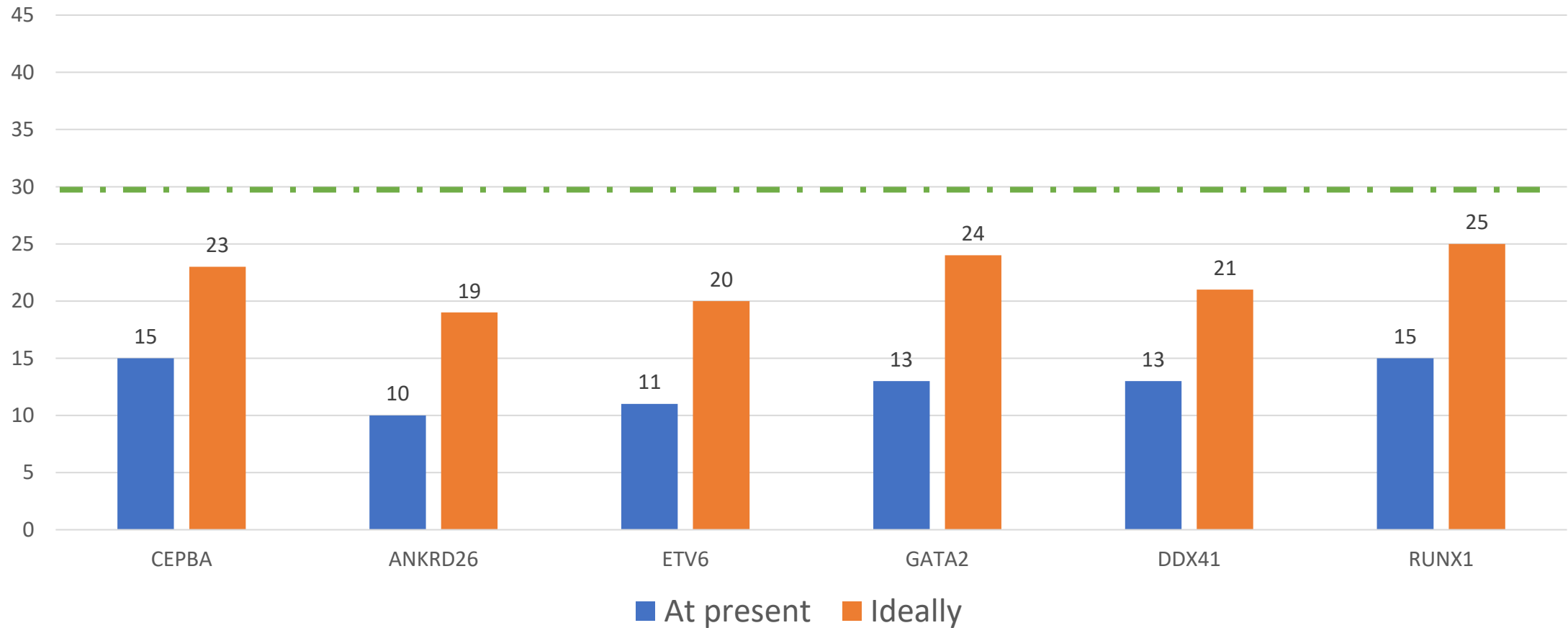
Consensus - 80% of respondents =30

If a germline variant is confirmed, predictive/cascade testing is offered to blood relatives



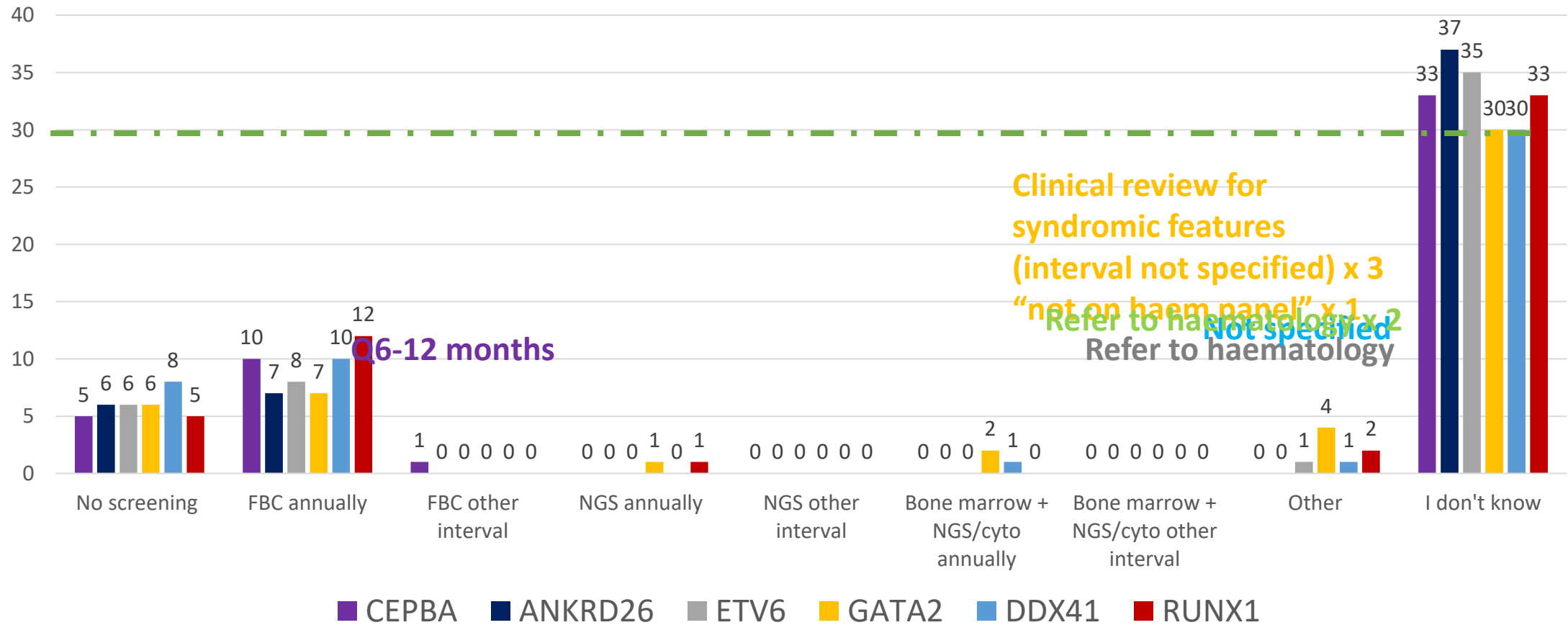
Consensus - 80% of respondents =30

Screening is offered to unaffected* carriers of pathogenic/likely pathogenic variants



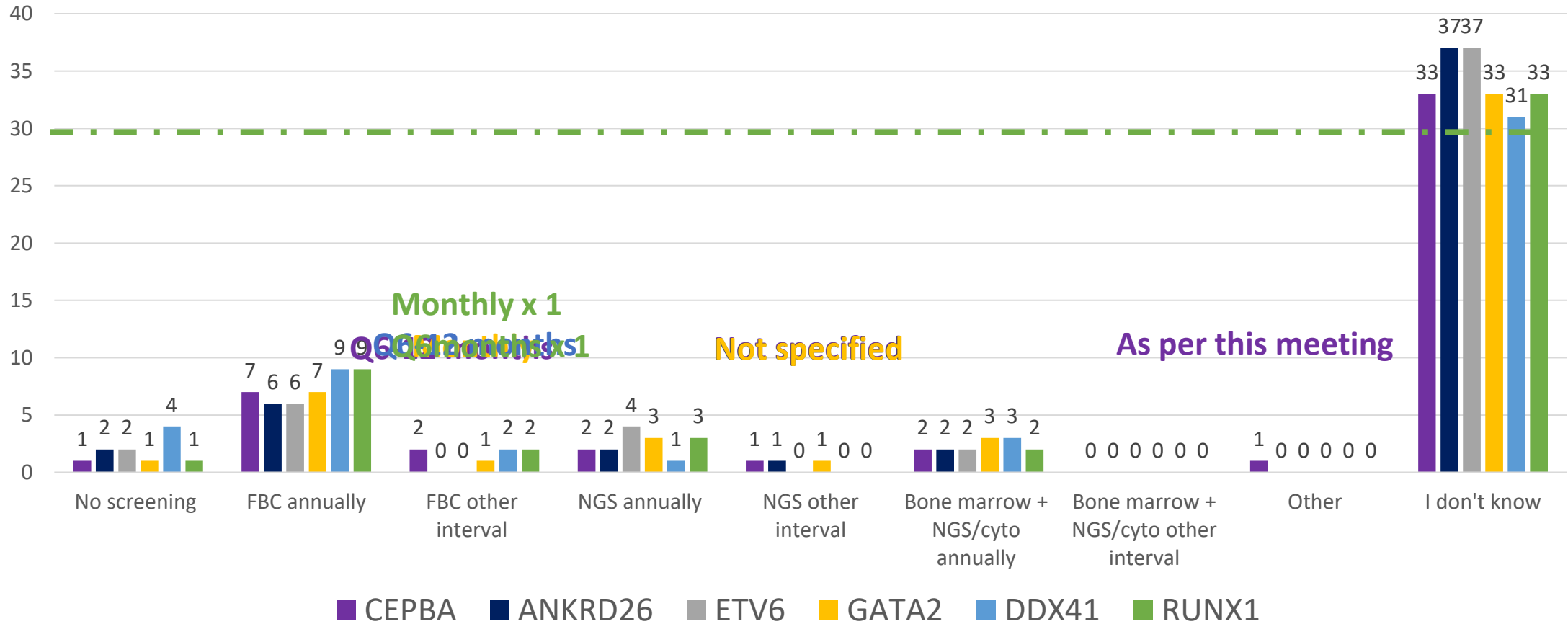
*no blood phenotype

AT PRESENT, what type of screening do you recommend for unaffected* carriers of likely pathogenic/pathogenic variants?



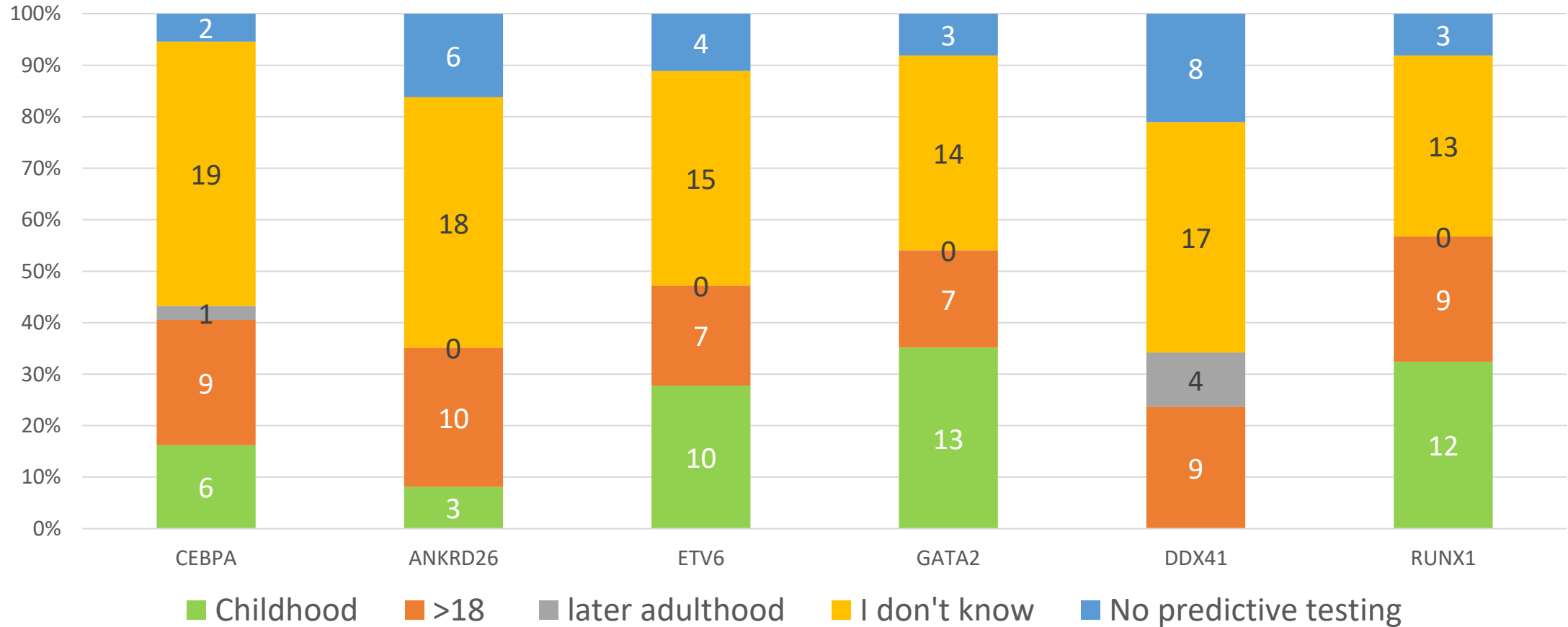
*no blood phenotype

Ideally, what type of screening do you recommend for unaffected* carriers of likely pathogenic/pathogenic variants?

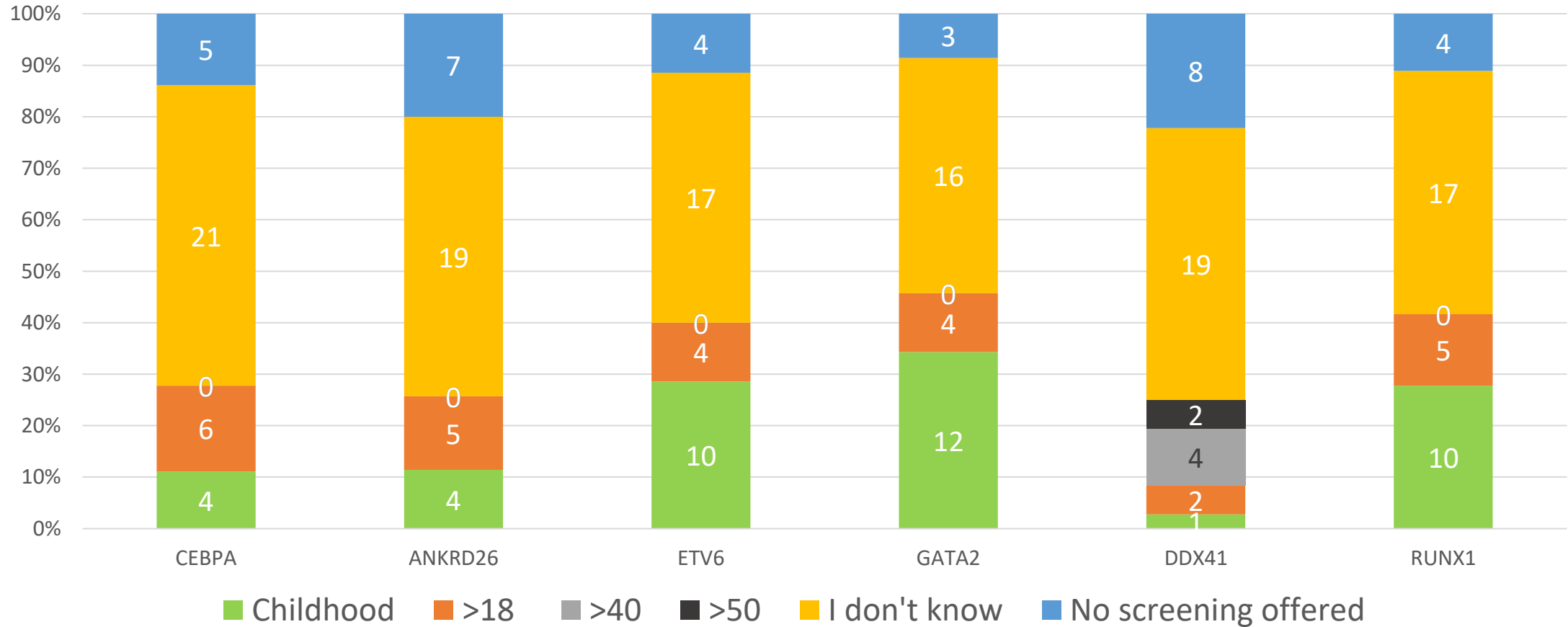


*no blood phenotype

From what age do you offer predictive testing?



From what age do you offer screening?



Summary

- General consensus – happy to offer confirmatory and predictive testing
- Agreement not yet reached about
 - Whether to offer screening
 - If screening offered, which type & frequency of screening should be offered
 - Age at which screening should start
 - Age at which predictive testing should be undertaken
- Additional things to consider
 - Who should be arranging predictive testing?
 - Who should be arranging follow up?
 - Assessments for non-haematological manifestations