Challenges looking after Children with Thalassaemia

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Paediatrician

SV-11 yr old girl

Born in SPH at term to Sri Lankan parents

Diagnosed with Thalassaemia major after new born screening test

Regular blood transfusions started age 3 months

Iron Chelation Therapy from age 18m

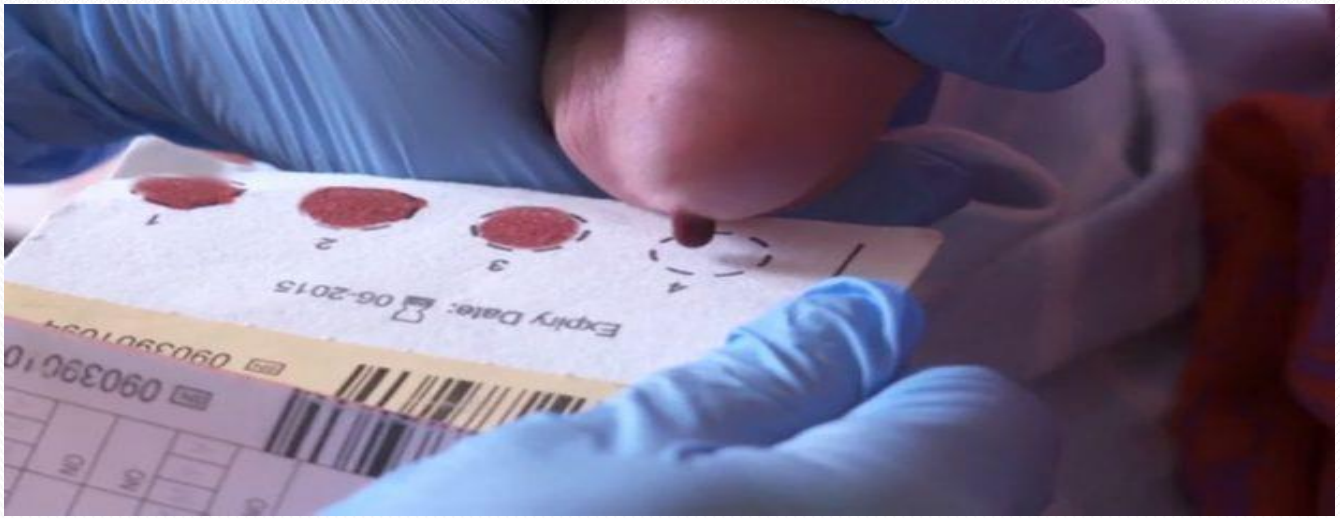
Regular monitoring, annual checks

PGD- failed twice (privately funded)

Plan for BMT discussed at an early age

New born Screening

- SCT: Mostly for SCD, CF and others
- Thalassaemia is also identified – referred to named paediatrician

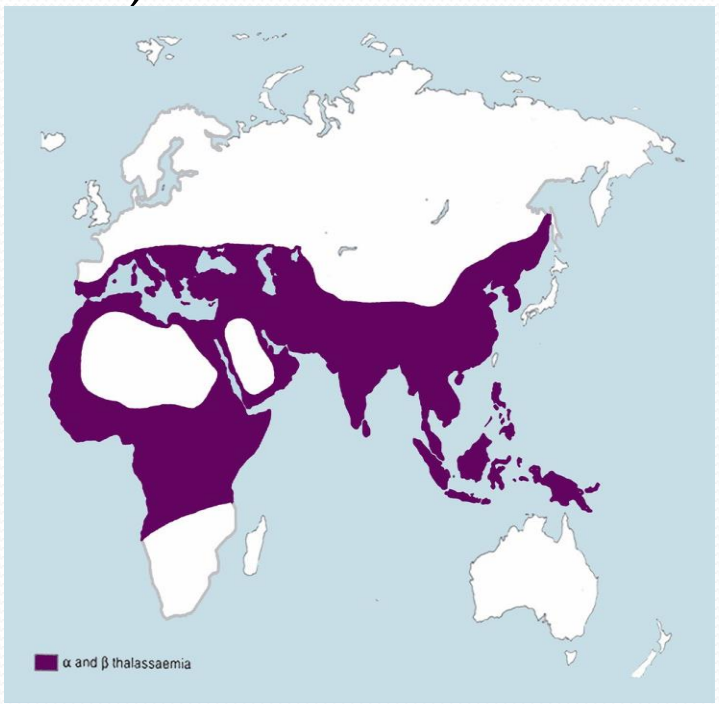


Genetics of Thalassaemia

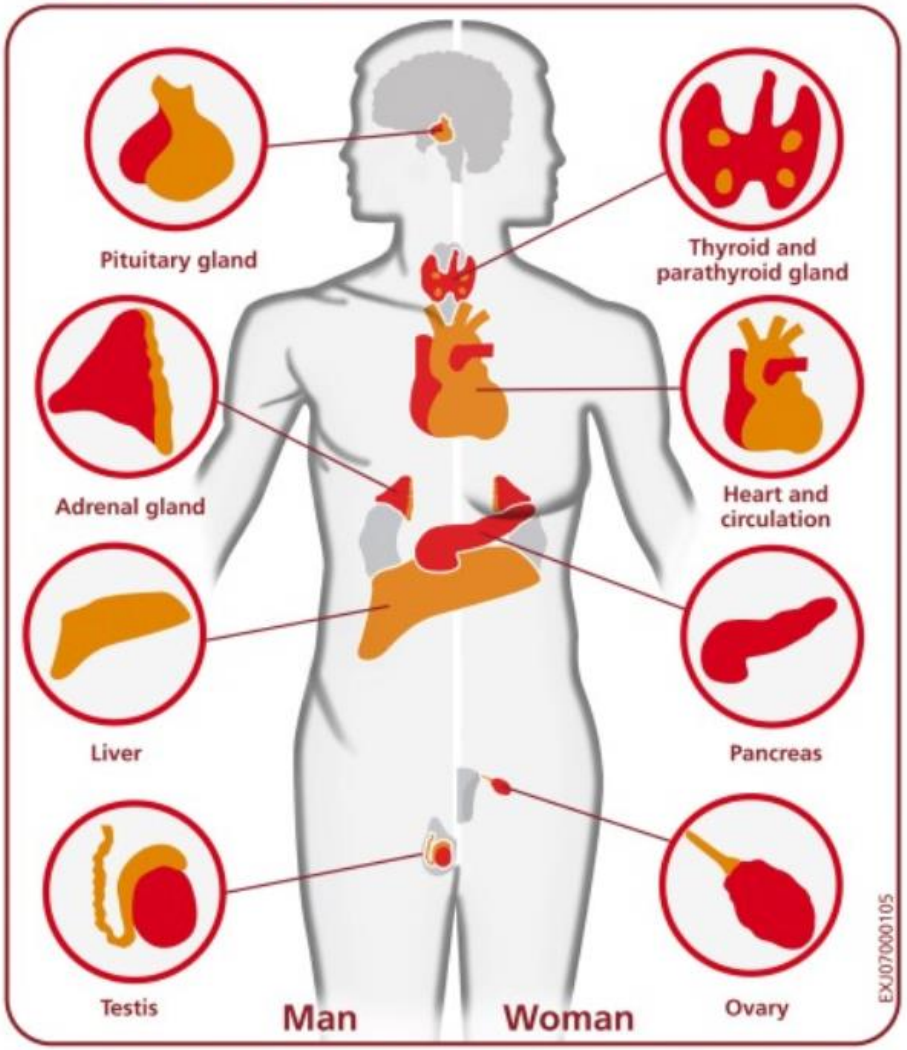
- AR condition – SE Asia, ME, Mediterranean regions – IVS 5 (C-G)
- Beta globin defect (quantitative)
- β0 or β+ or β++
- Other beta chain abn:

  Hb C, H, E, D, O
- 20-30 babies born

  in the UK



Organs that may be affected by iron overload



Toxic iron builds up across the body and can cause serious damage to vital organs, including the heart and liver.

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Treatment Options

- Regular transfusion therapy
  - To treat anaemia, prevent complications
- Extended lab tests for these patients
- Transfusions:

  Pre transfusion: 90-100

  Post transfusion: 140

  Annual: < 200ml/kg

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Iron Chelation Therapy (ICT)

- Main drugs:

  Desferrioxamine (DFO):

  Deferiprone (DFP):

  Deferasirox (DFX):

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Monitoring & F ups

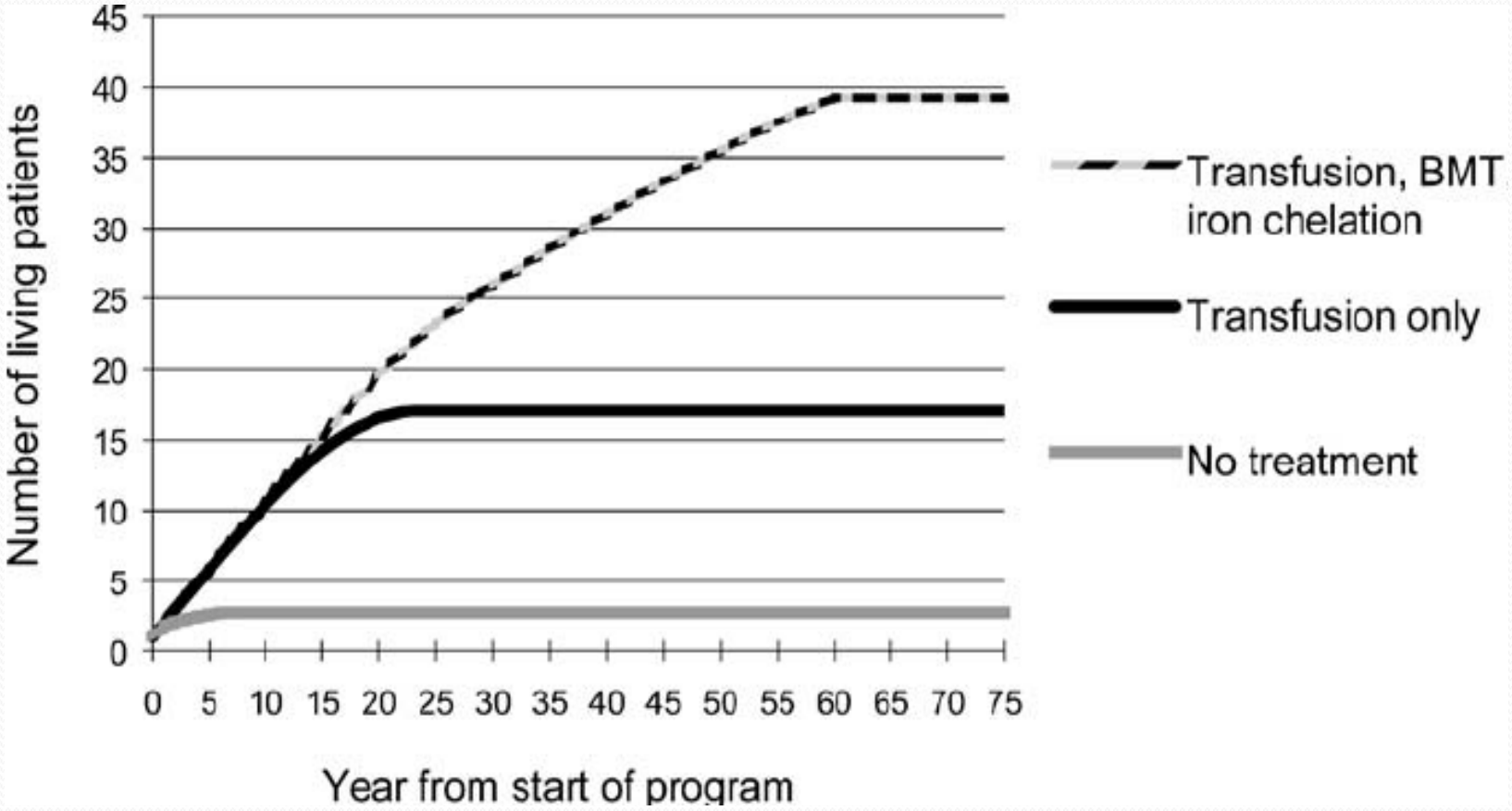
- Mainly related to iron overload or drug toxicity

IRON OVERLOAD:

S. Ferritin- crude, unreliable

LIC: liver biopsy, MRI (> 7mg/g dw= high morbidity).

HEART MRI T2*: <10msec=high risk for HF


Number of living patients
45
40
35
30
25
20
15
10
5
0
0 5 10 15 20 25 30 35 40 45 50 55 60 65 70 75
Year from start of program
Transfusion, BMT iron chelation
Transfusion only
No treatment

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Preimplantation Genetic Diagnosis

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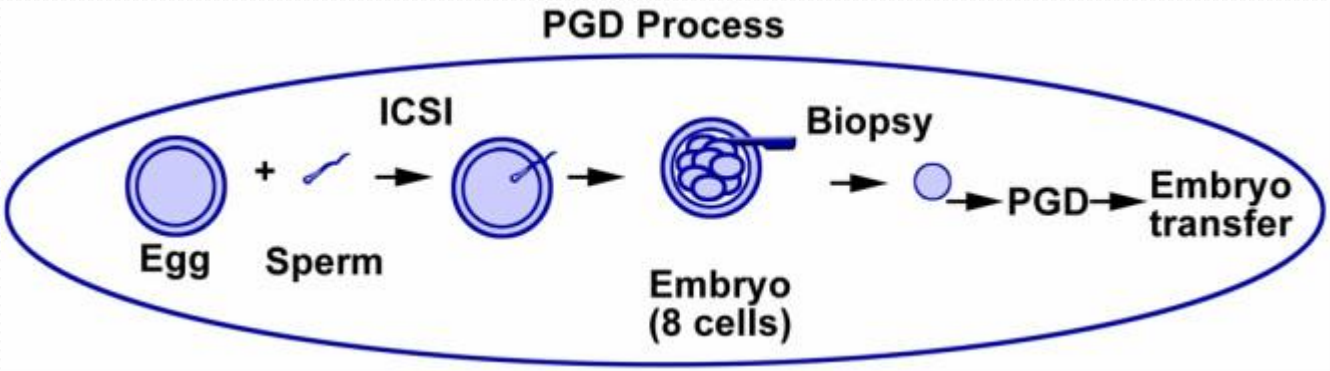

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Haematopoietic Stem Cell Transplant

- HLA matched sibling donor: cure 80-90%
- High resolution tissue typing techniques:
- Haplo-identical HSCT:
- Cord blood SCT: easy with comparable results

Future- Gene Therapy

Small study: journal of human gene therapy