

## Presentation

### Before first transfusion:

- FBC and reticulocytes anaemia and low retics
- eADA >100 nmol/mg Hb/h
- HPLC >1% or adjusted for age
- Serology for parvovirus, hepatitis B, hepatitis C and HIV negative

### Diagnosis:

- Bone marrow biopsy:
  - aspirate and trephine absence or reduction beyond proerythroblasts
  - cytogenetic analysis and FISH negative
  - parvovirus PCR negative
- Mutation analysis

- Examine for skeletal abnormalities: palate, limbs, spine and scapula
- Testicles
- USS abdomen
- echocardiogramme
- hearing test
- ophthalmology review

Hepatitis B vaccine

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graph TD; A[Hepatitis B vaccine] --> B[Transfusions minimum to 12 months]; B --> C[Investigate immune system: <br/>• lymphocyte subsets <br/>• immunoglobulins <br/>• Immunoglobulin subclasses <br/>• responses to antibodies]; C --> D[MMR <br/>Chickenpox vaccine]; D --> E[trial of prednisolone 2 mg/kg for four weeks];
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Transfusions minimum to 12 months

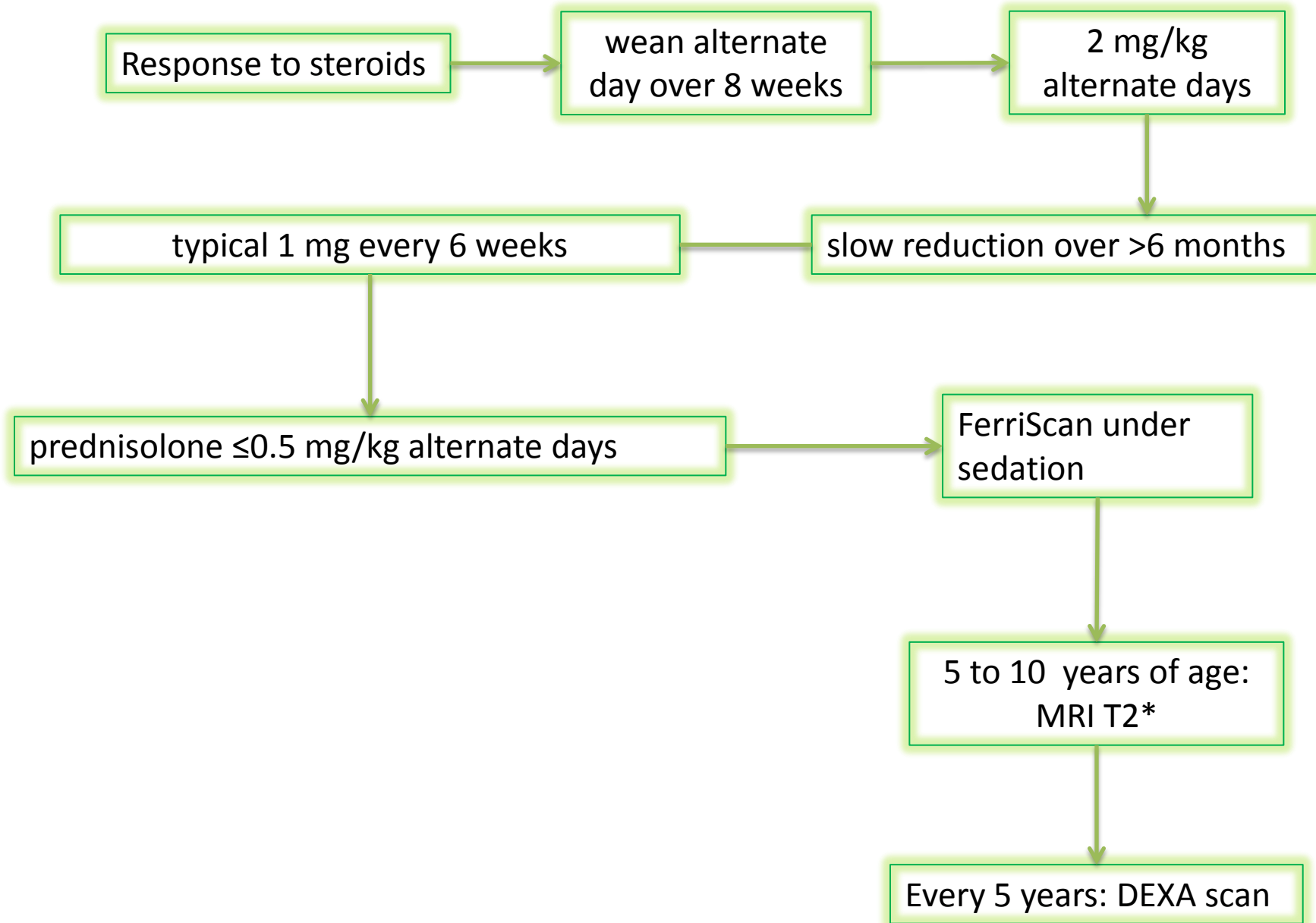
Investigate immune system:

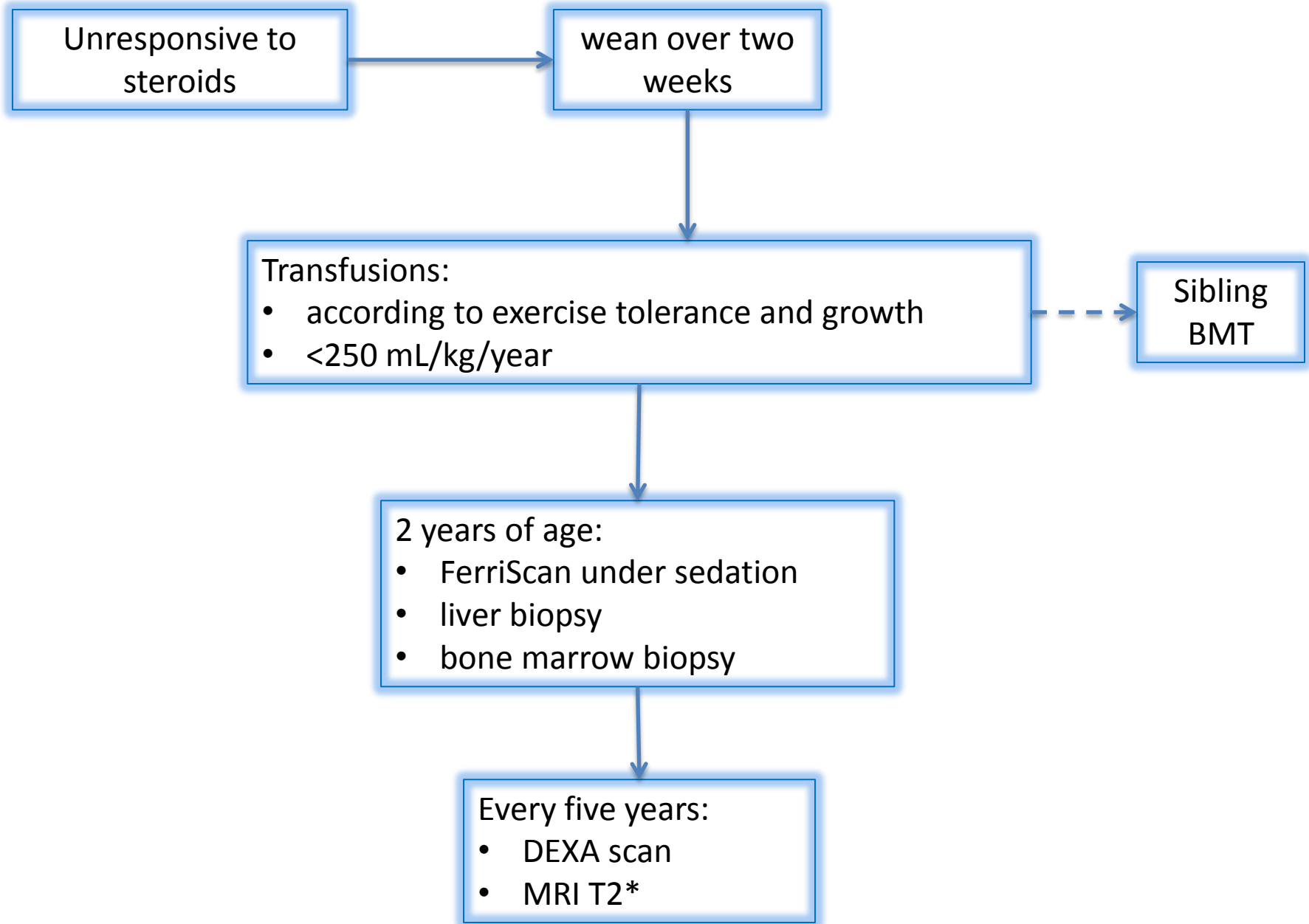
- lymphocyte subsets
- immunoglobulins
- Immunoglobulin subclasses
- responses to antibodies

MMR

Chickenpox vaccine

trial of prednisolone 2 mg/kg for four weeks





bone marrow biopsy if cytopenia

monitor film  
vitamin D

yearly endocrinology review from 10 years of age  
until end of pubertal development