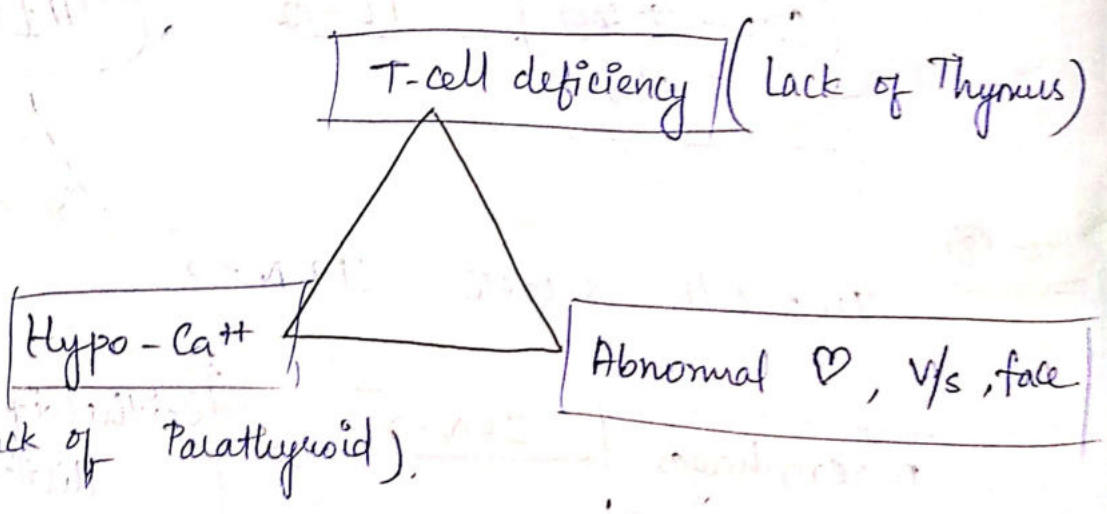
Synopsis:

(i) Digeorge Syndrome
(ii) SCID.
(iii) X-linked Agammaglobulinemia
(iv). CVID.
(v) IgA deficiency (GIT - celiac ds.)
(vi) Hyper IgM syndrome
(vii) Wiskott - Aldrich syndrome.
(viii) Complement Deficiences.

(i) Digeorge syndrome.

* Developmental failure of 3rd & 4th pharyngeal pouches.
* 22q11 microdeletion

Characteristics



Defective Cell mediated as well as Humoral immunity.

ETIOLOGIES (3)

- Cytokine ® defects
  - ⇓
  - cytokines are essential for ⊕ of T-cell, B-cell so....
- Adenosine Deaminase Deficiency
  - ⇓
  - Without Deaminatn, adenosine & deoxy Adenosine builds up in lymphocytes
  - ⇓
  - Toxic to lymphocytes
  - ↓
  - Immunodeficiency
- MHC class II deficiency
  - CD4+ (4×2=8) T-cell.
  - ⇓
  - Helps both CD8+ T- & B-cell
  - ⇓ That why
  - Both CMI & Humoral immunity is aff.

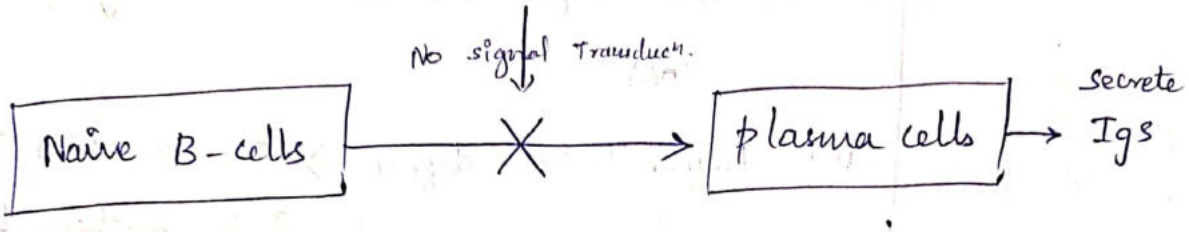
Characteristics:

- Susceptibility to
  - * fungal.
  - * viral
  - * bacterial
  - * protozoal infecn.
- ↑ risk to oppurtunistic infecn & Live Vaccines.

Rx:
- sterile isolation.
- Stem cell transplant (Hematopoietic stem cell. → T-Cell, B-cell)

Ig in blood } Complete lack of Ig in blood

* Due to disordered B-cell maturatn.



* X-linked mutatn in BTK - Bruton Tyrosine kinase ⇒ "signalling molecules."

Presentation: After ⑥ months of life.

Recurrent infecn of

Bacteria → without IgG → No opsonizatn.

Entero virus

Giardia

* affects mucosa of GIT
* ∵ pt. doit have IgA (mucosal Ig)

Why After ⑥ months? Bcoz upto ⑥ months, fetus has mom's Igs..

Caution:
Live vaccines (eg. polio) should be avoided.

CVID = Common variable ... g

Low Immuno globulin in blood

↓ why?

(i) B-cell defect ⇒ due to which Ig synthesis ↓

(OR)

(ii) Helper T-cell defect ⇒ due to which IL-4 & 5 synthesis ↓

so B-cells → plasma cells

Presentation:

* ↑ risk of infections in late childhood

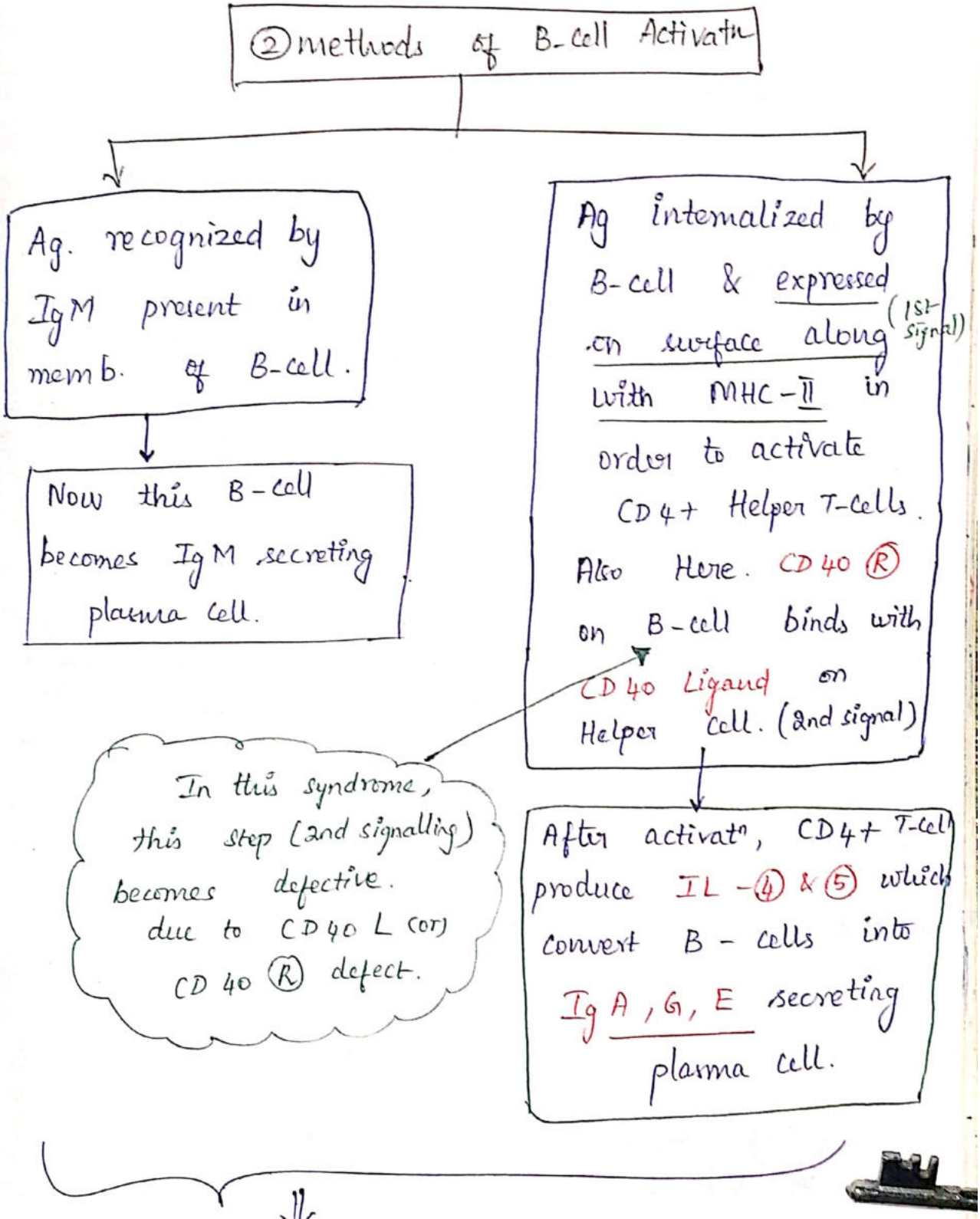
- Bacteria
- Entero Virus
- Giardia

* ↑ risk of (i) Auto immune ds:
(ii) Lymphoma.

(v) Ig A deficiency - m/c Ig deficiency

* Low serum & mucosal Ig A.
* ↑ risk of mucosal Inf. especially "viral".
* In Celiac ds, pt. presents with Ig A def.

* Due to Mutated CD40 L (or) CD40 (R).



⇓

in this ds, only 1st step is normal, 2nd method is abnormal/defective, so after 1st method, Large cunt. of IgM secreting plasma cells will be there → Hyper IgM Syndrome

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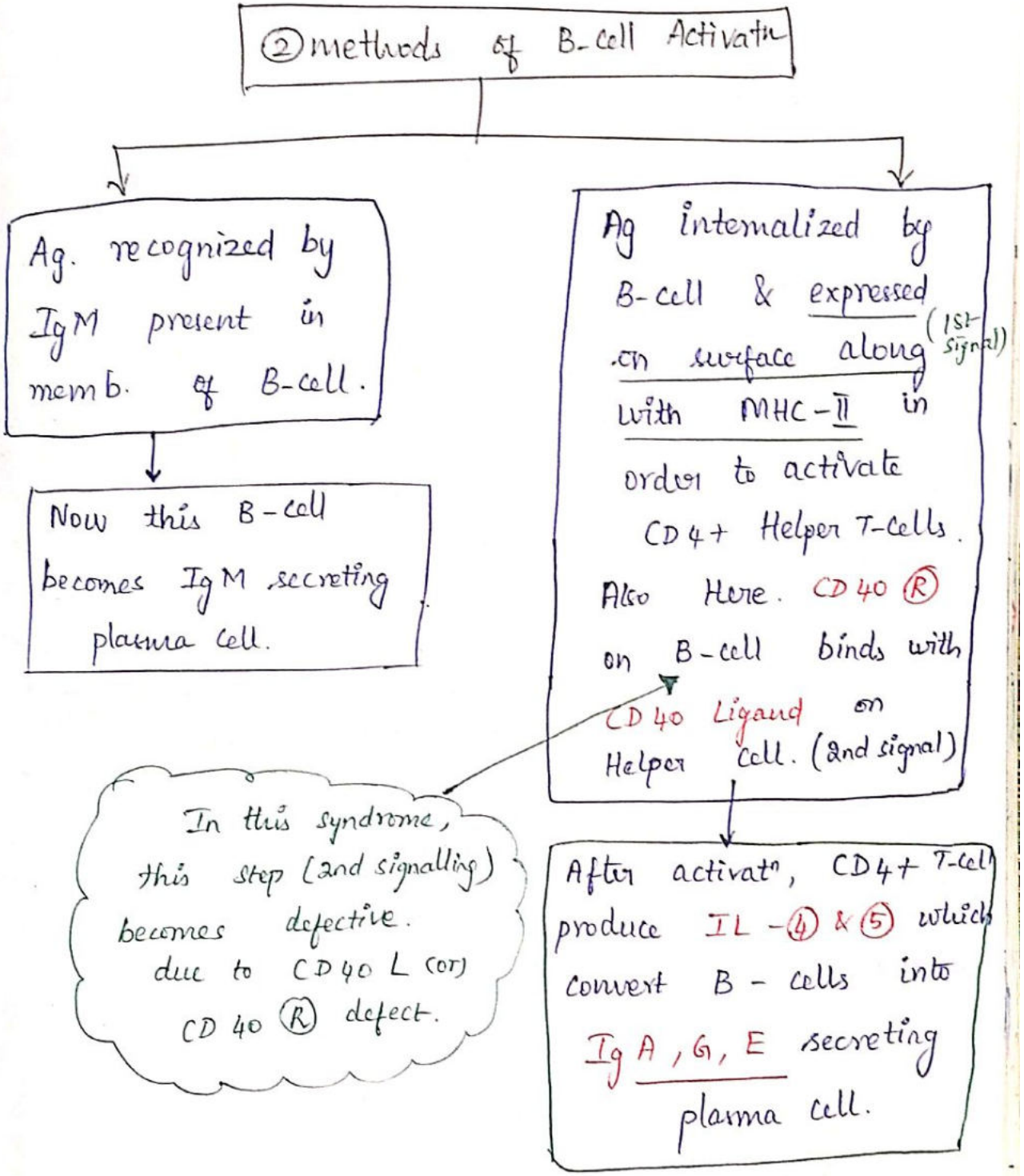

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(2nd signal in 2nd method of B cell activation is not delivered to T-cells)

↓

cytokines (IL-4, 5) necessary for Ig class switching not produced

↓

IgA, IgG, IgE are low.

low → Mucosal infecn.

low → recurrent pyogenic infecn (bcoz IgG is an opsonin).

(vii) Wiskott - Aldrich Syndrome (WAS).

Triad

- Thrombocytopenia → causes petechia in skin & mucosa.
- Eczema (skin rash)
- Recurrent Infecn. (due to defective Humoral as well as Cell med. imm.)

Mutation: WASP gene (X-linked).

↳ (Wiskott Aldrich ~~Syndrome~~ Protein)

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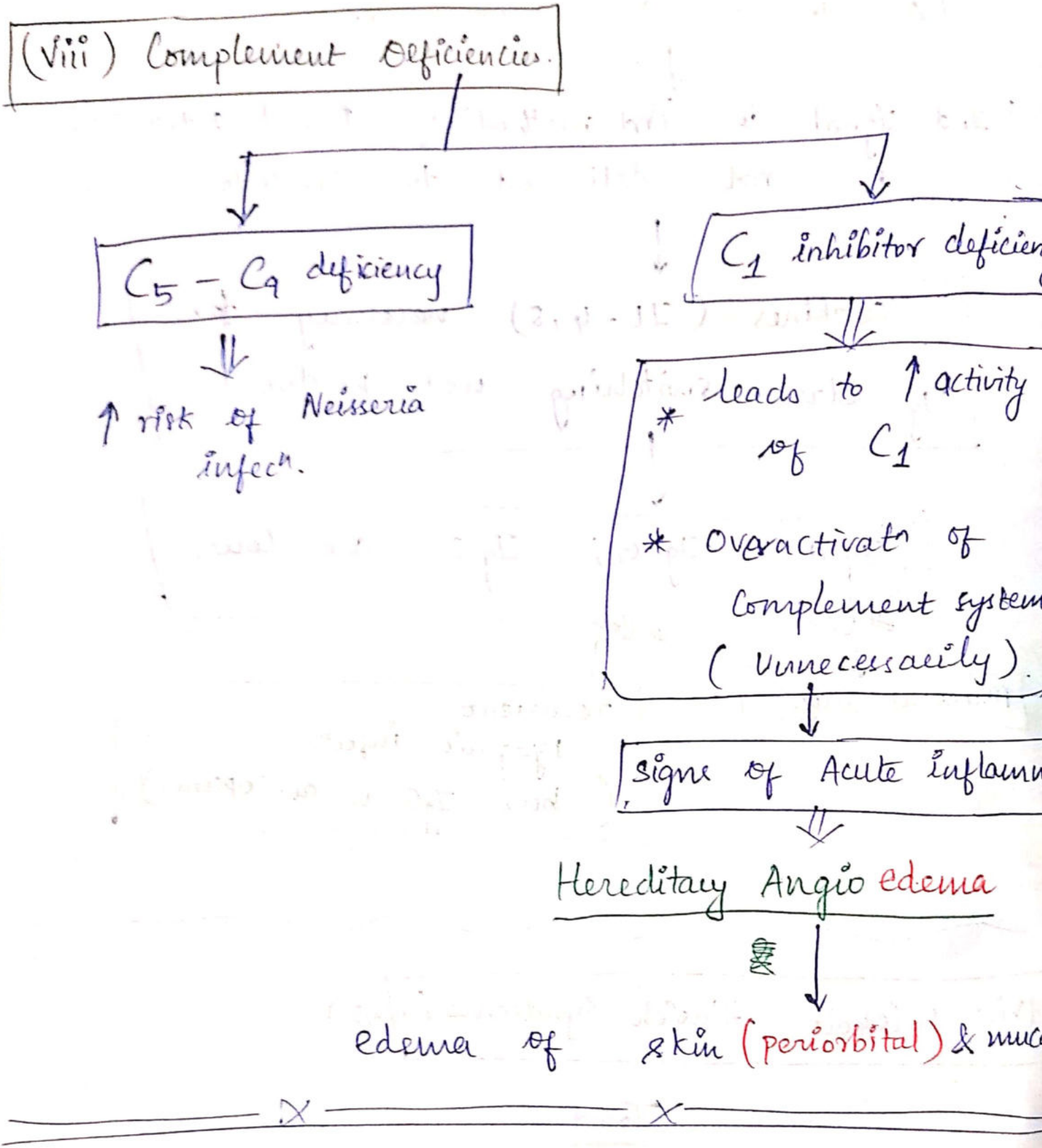

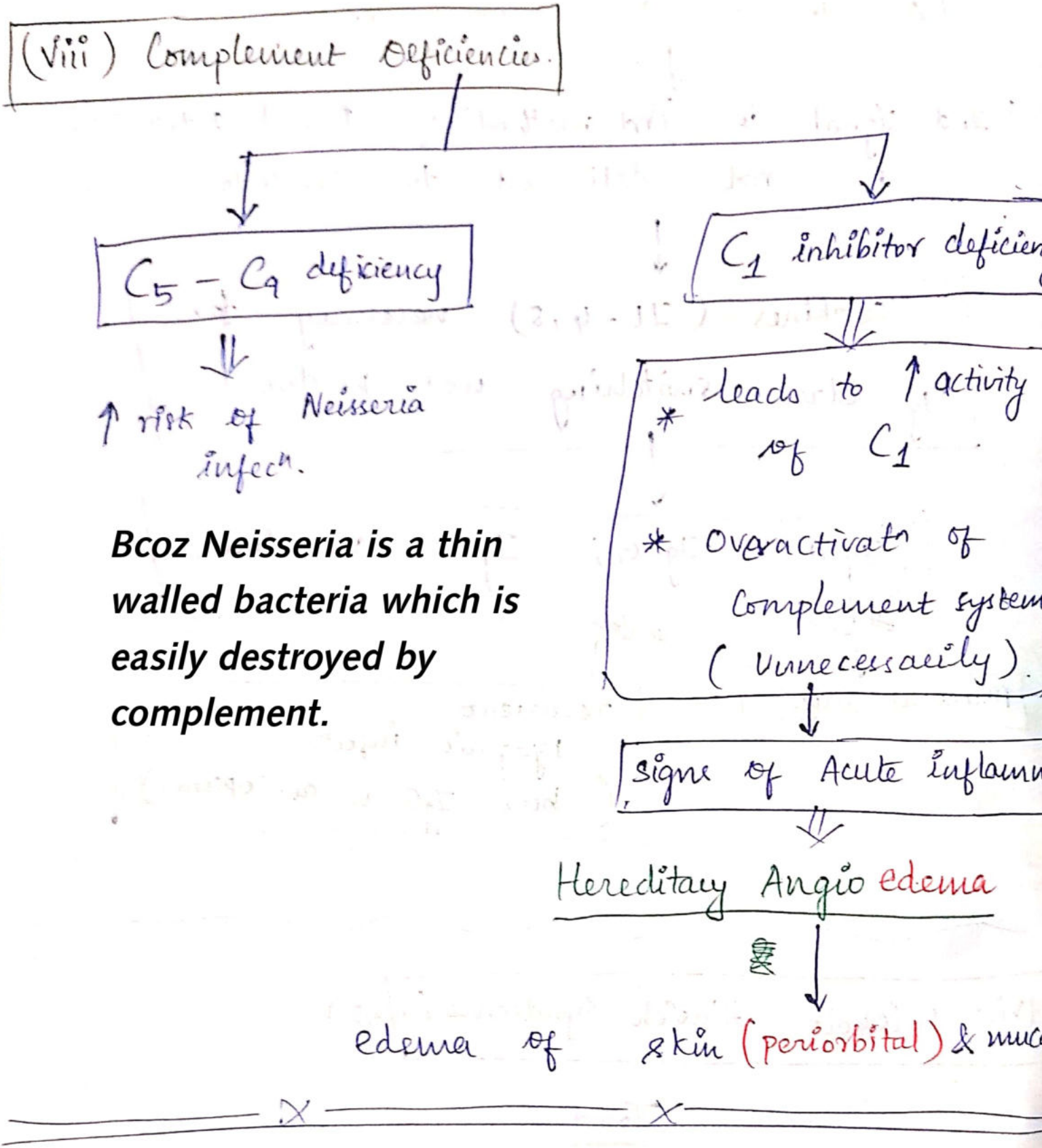

Bcoz Neisseria is a thin walled bacteria which is easily destroyed by complement.