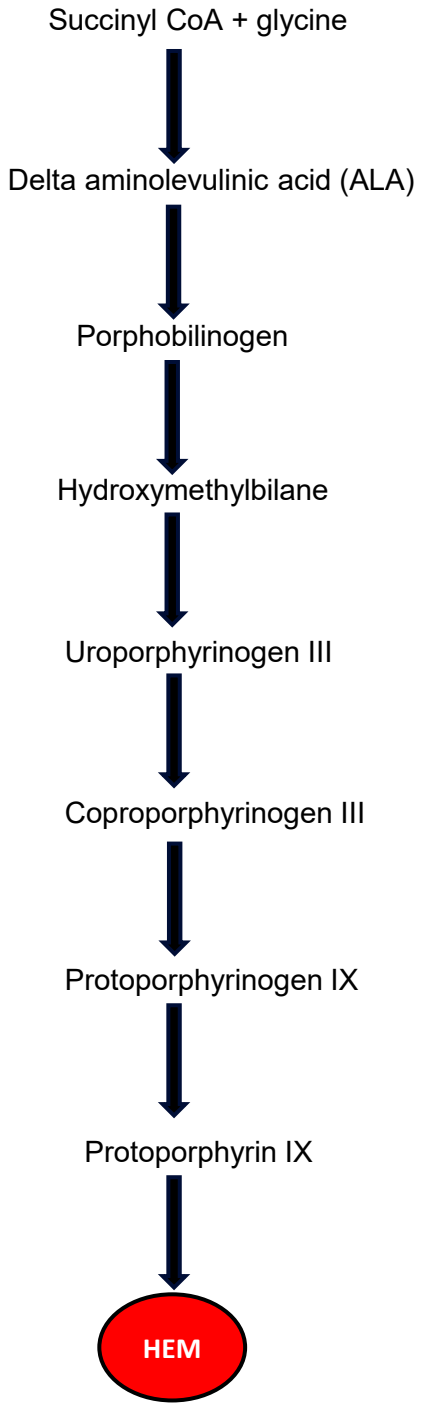
Hvorfor får man utløst sykdommen?

2. og 3. november 2026

Øyvind Skadberg
Overlege
Nasjonalt senter for sjeldne diagnoser, enhet porfyrisykdommer (Napos)
Haukeland universitetssykehus

NASJONALT SENTER FOR
SJELDNE DIAGNOSER

HEMSYNTESEN

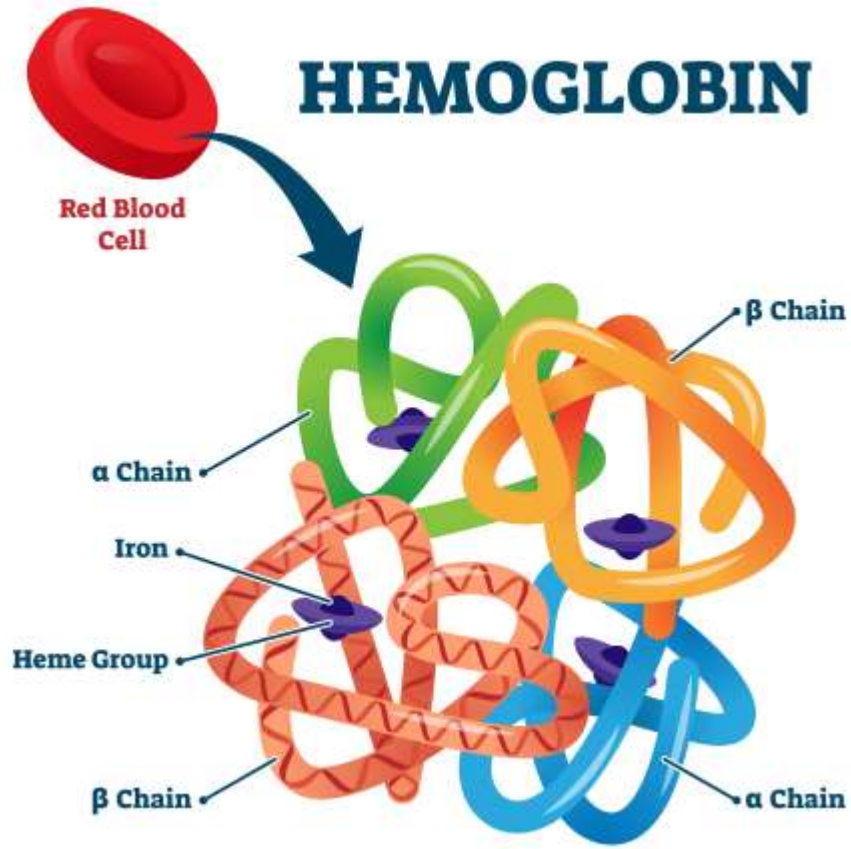


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SJELDNE DIAGNOSER

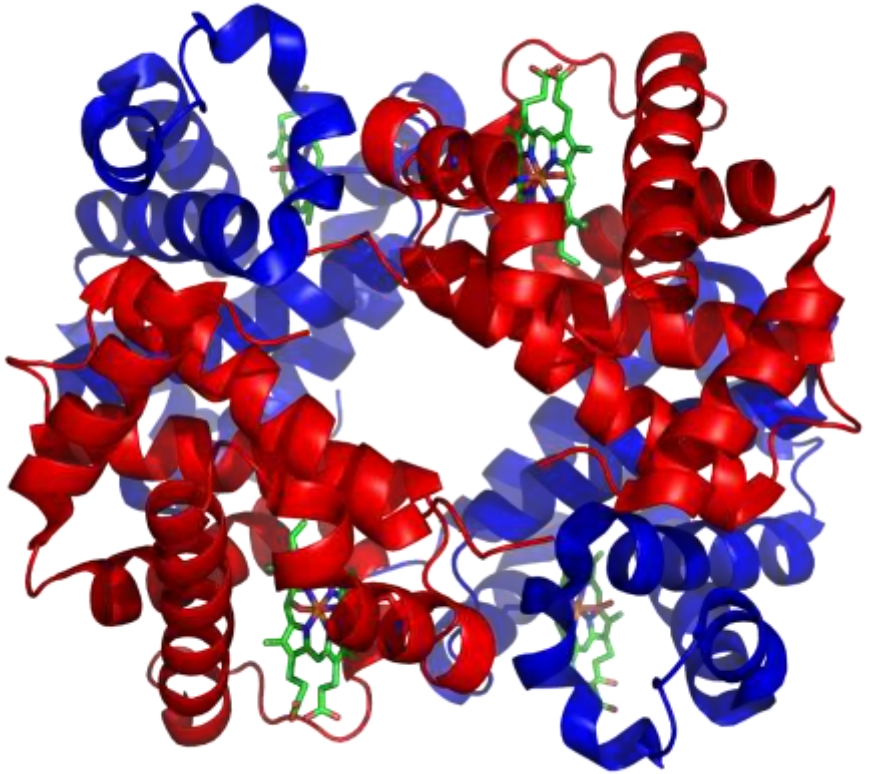
Figur: Solveig B. Glad, NAPOS

Vi må snakke litt om vanskelige ord

- Porfyriner
- Enzymer
- Samlebånd
- Leversykdom


HEMOGLOBIN
Red Blood Cell
β Chain
α Chain
Iron
Heme Group
β Chain
α Chain

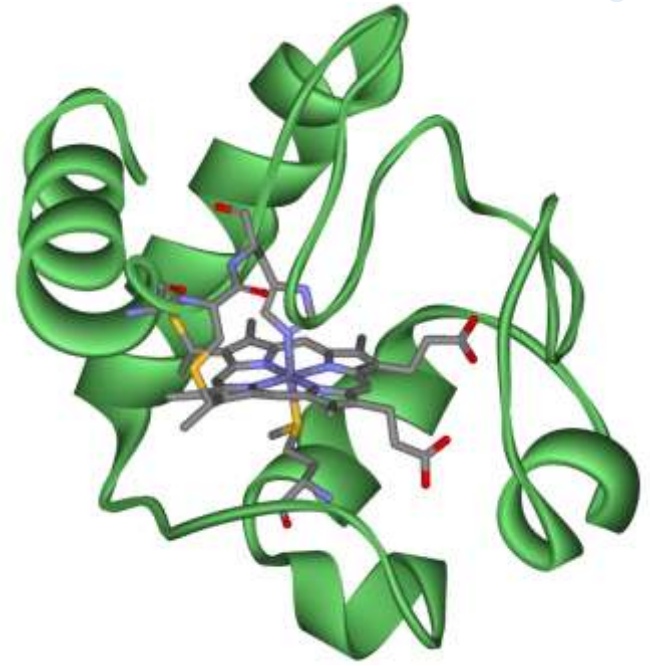
Hemoglobin



By Zephyris at the English-language Wikipedia, CC BY-SA 3.0

«Heme» - ikke bare i røde blodceller

- Heme lages i alle celler som har cellekjerne
- I leveren brukes heme til å lage proteiner, (enzymer, cytokromer)
- PCT er en «leversykdom»

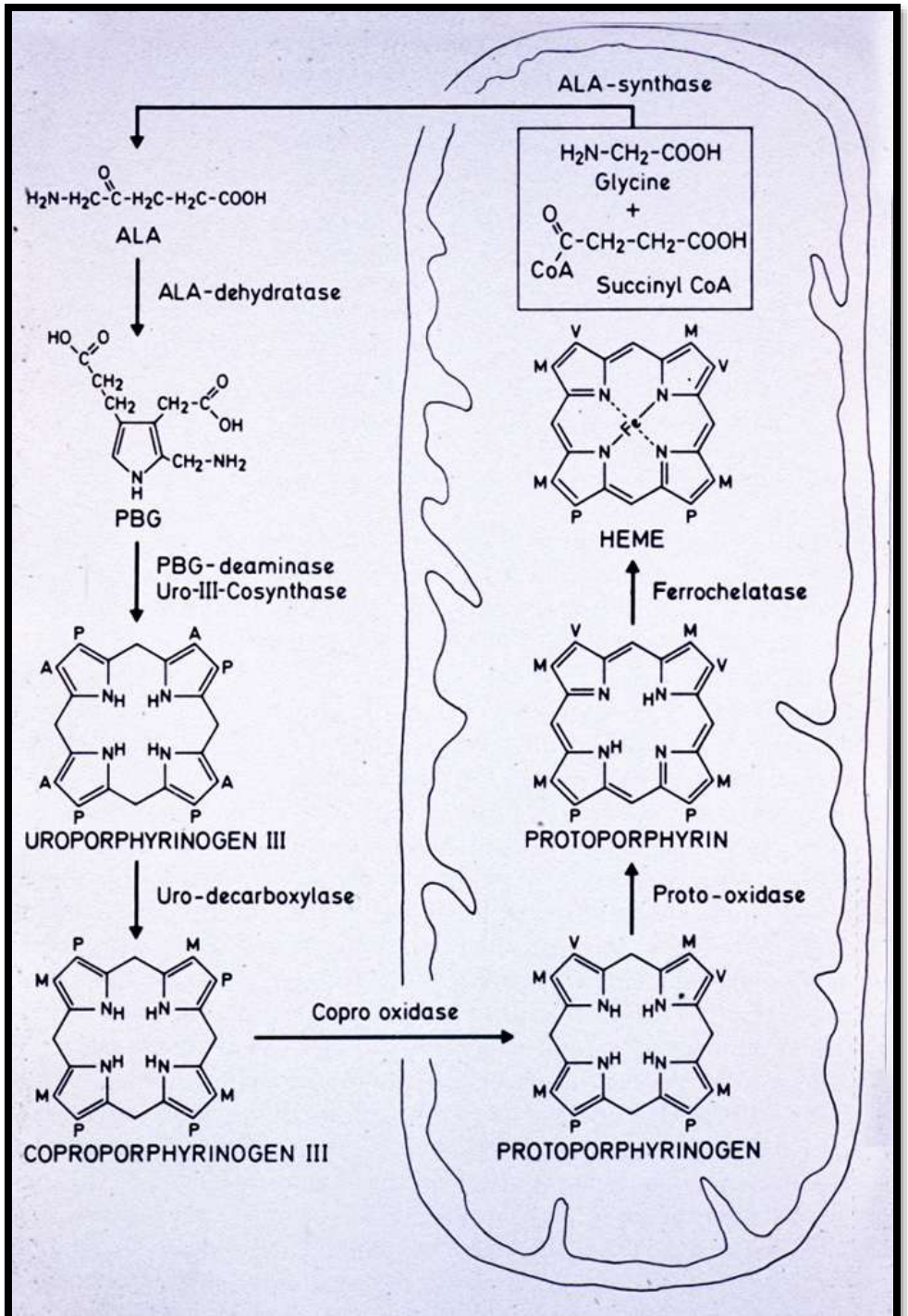


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https://en.wikipedia.org/wiki/Cytochrome

5. Enzym i hemesyntesen

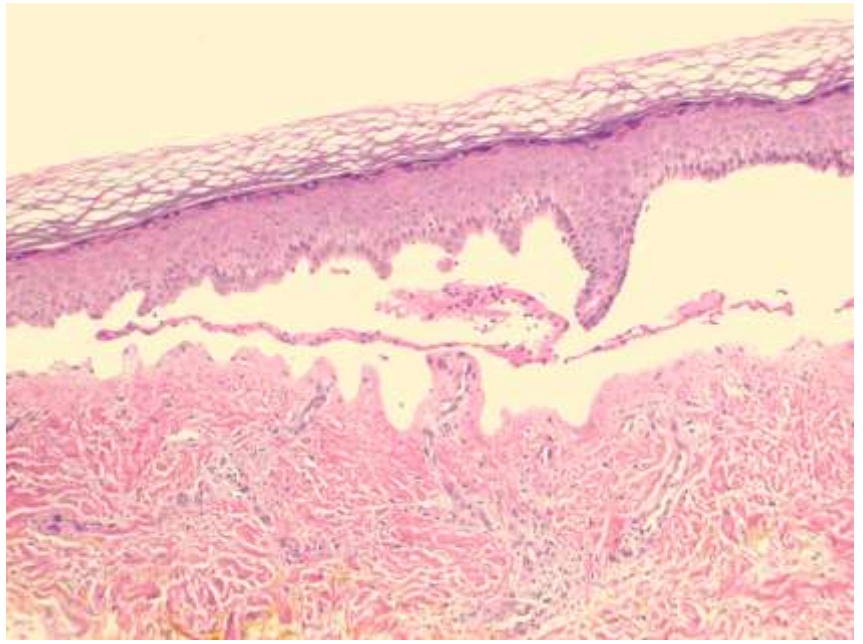
URO-d

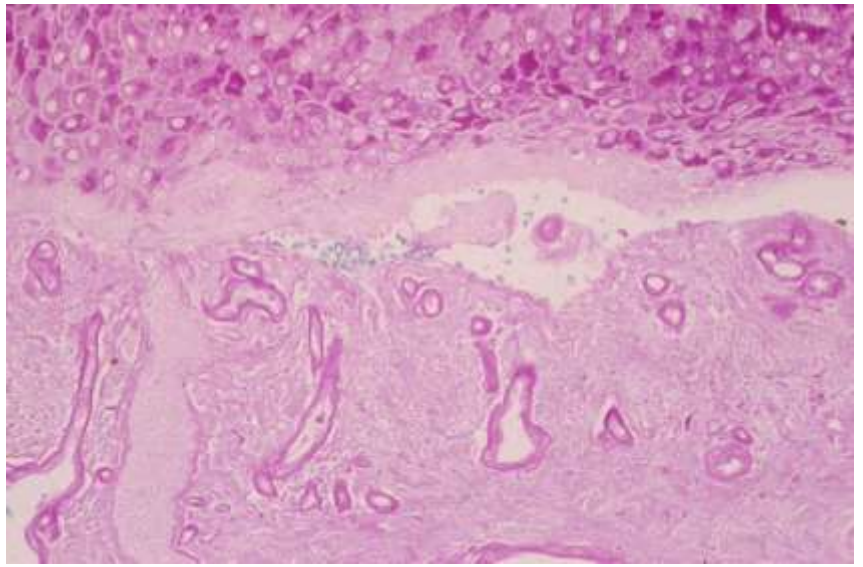


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SJELDNE DIAGNOSER

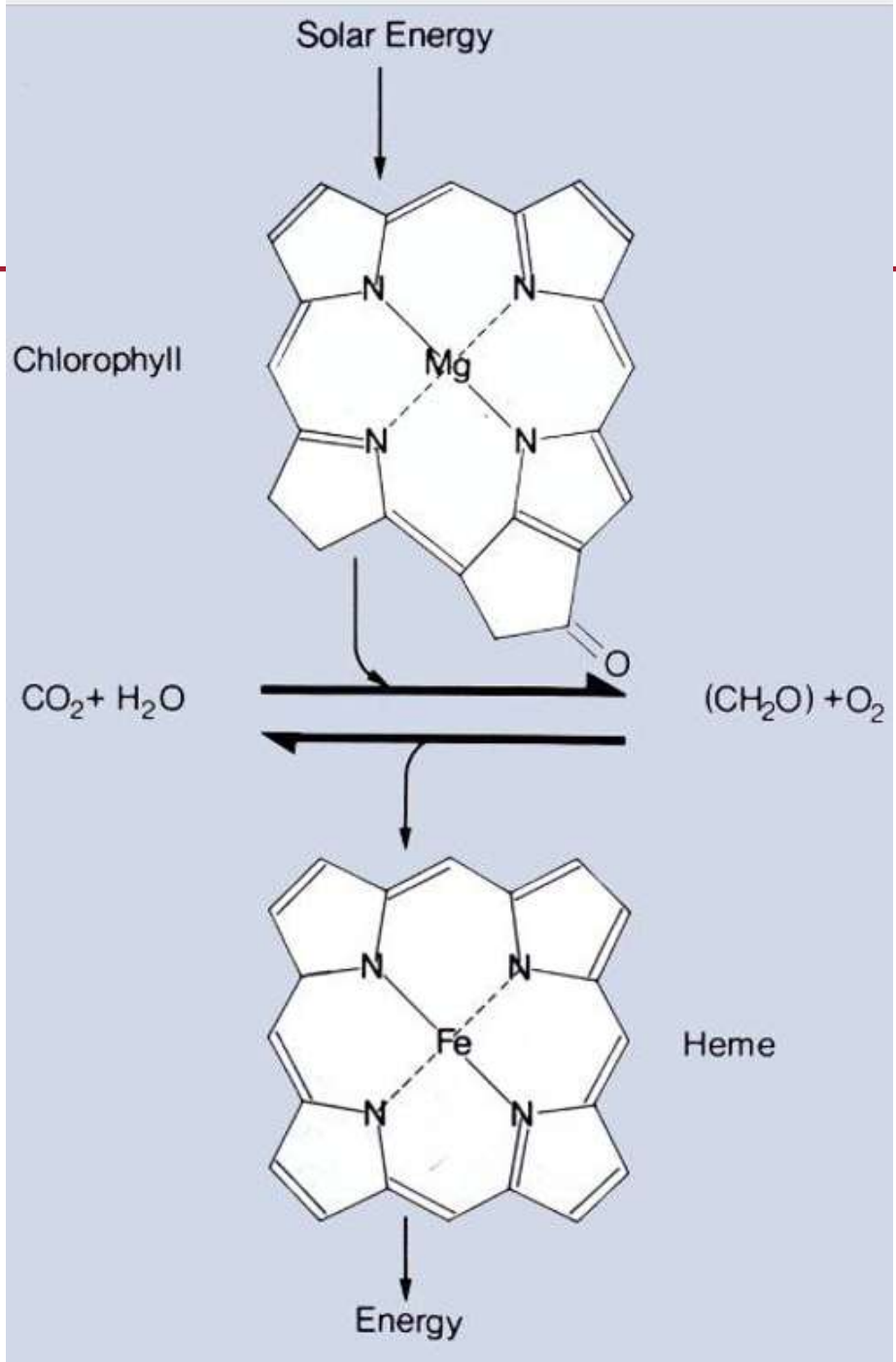
Hud med «blemmer» og sår



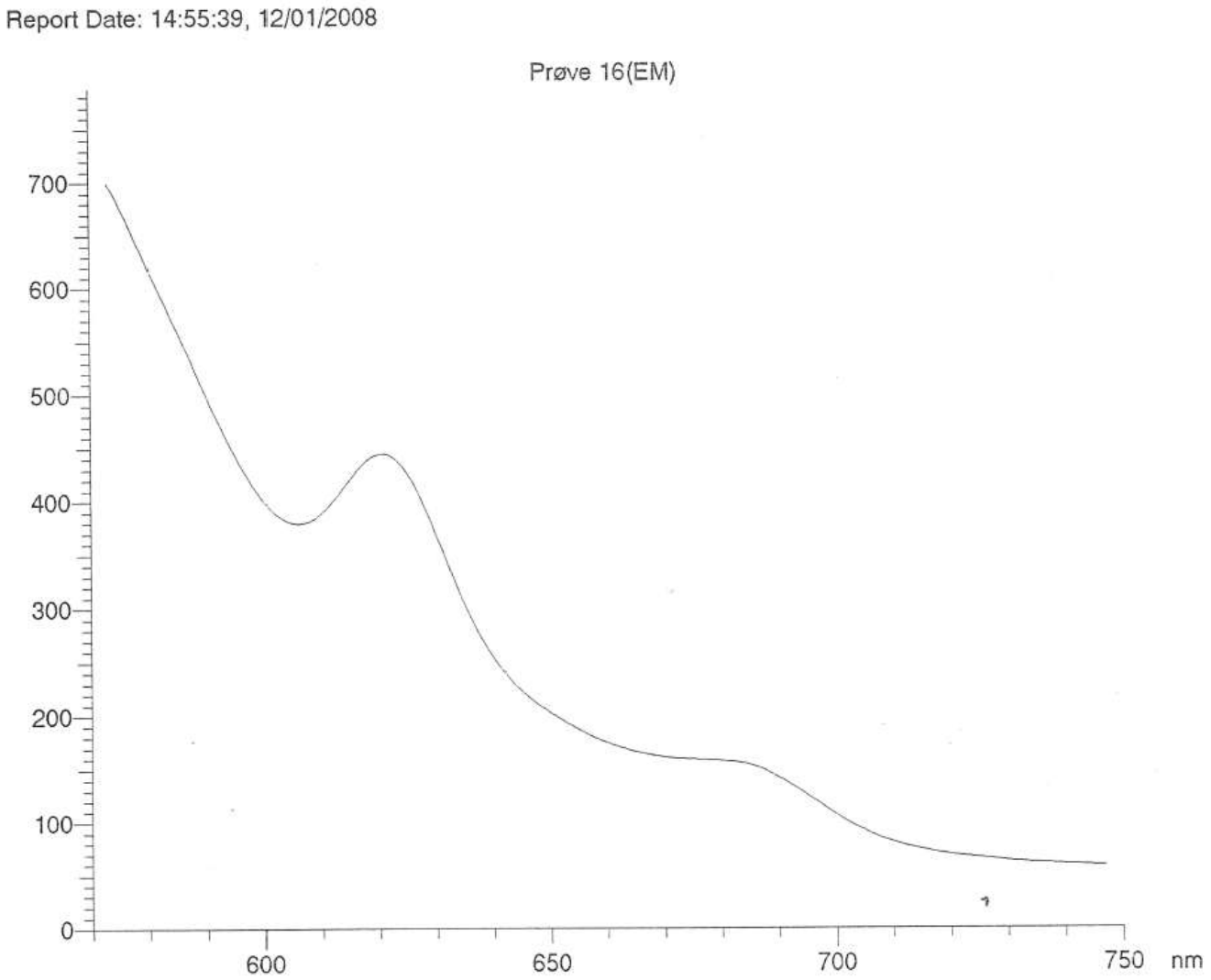




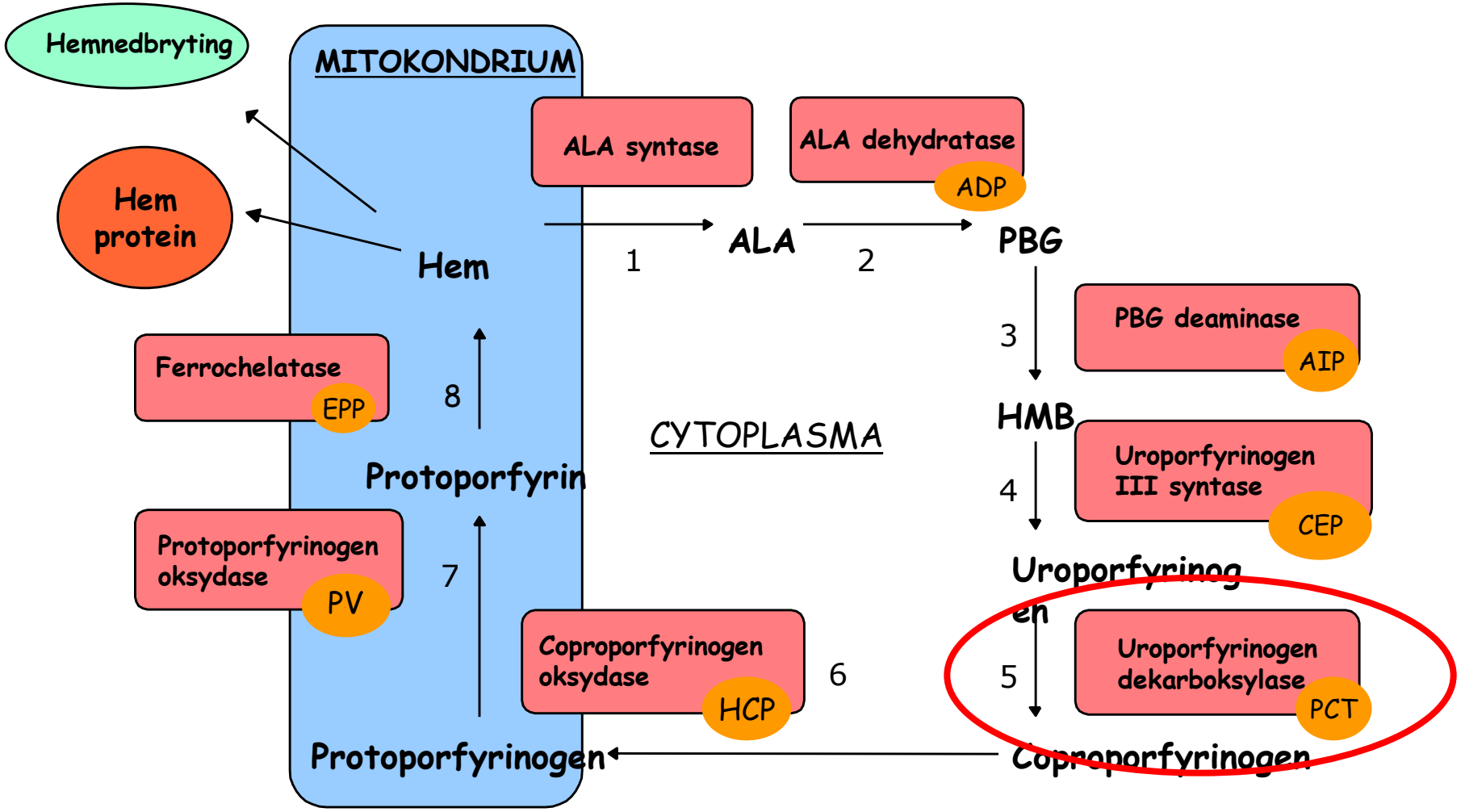
Hvorfor skades huden ?

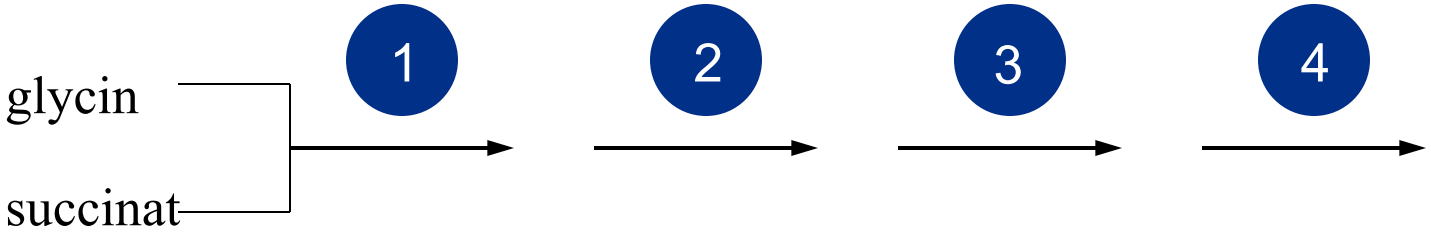


Positiv plasmaporfyrin analyse

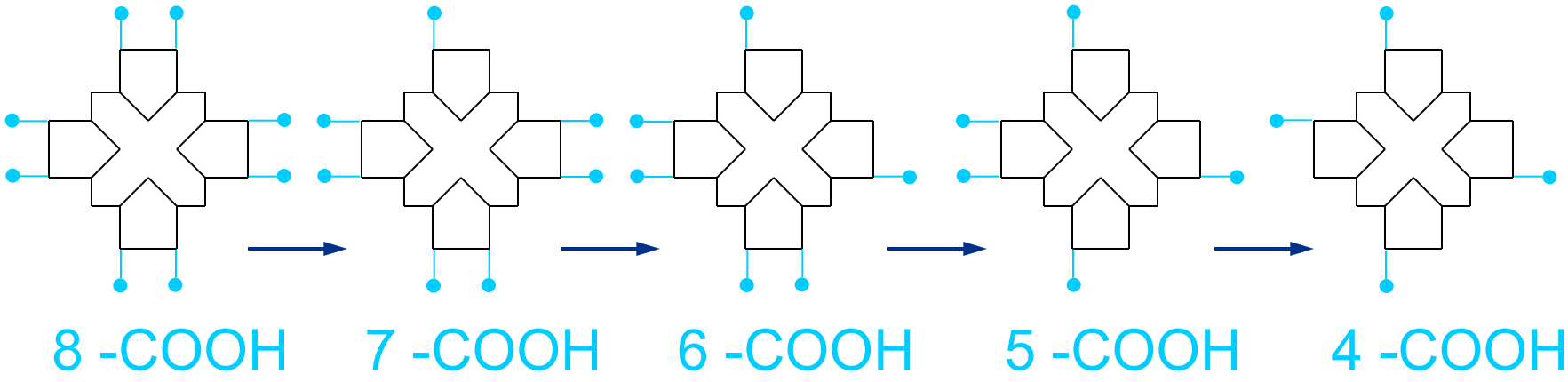


Hem biosyntese og arvelige enzymdefekter





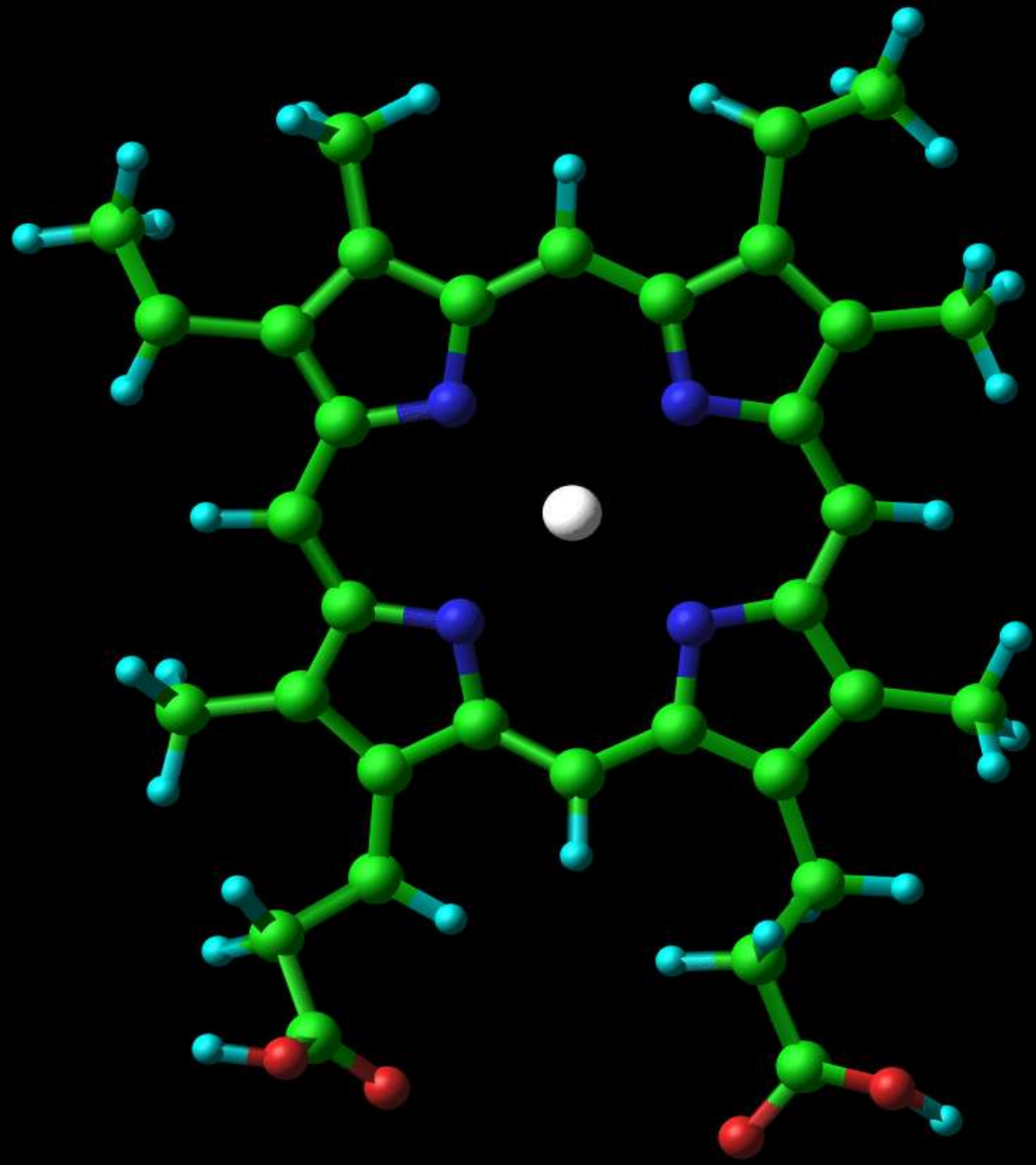
5 *Uroporfyrinogen dekarboksylase (Uro-D)*



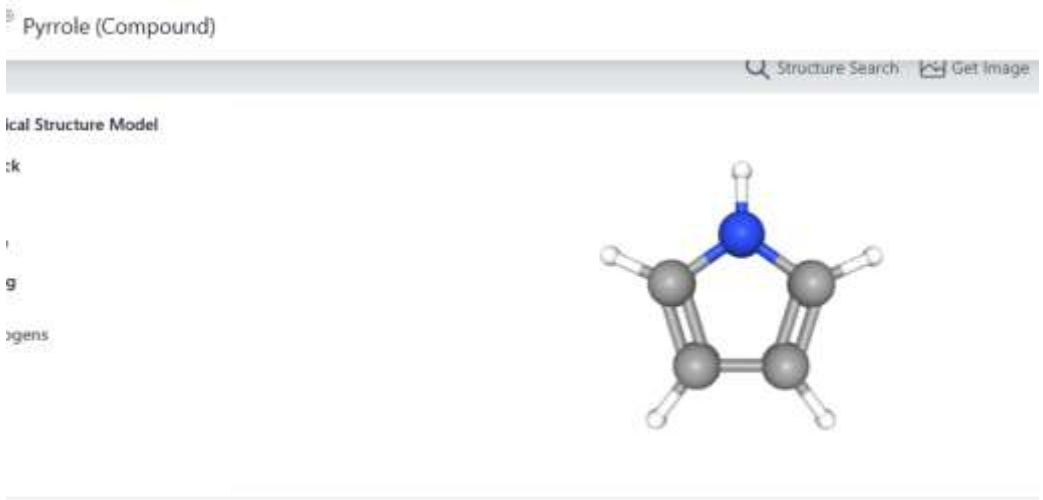
Uroporfyrinogen dekarboksylase (Uro-D)

- Økt mengde polykarboksylerte porfyriner
- Vannløselige, - utskilles i urinen

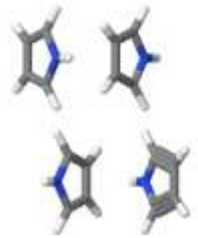
U-Porfyriner (Totalt)	↑↑
U-Uroporfyrin (8 -COOH)	↑↑
U- Heptaporfyrin (7 –COOH)	↑
U-Hexaporfyrin (6 –COOH)	↑
U-Pentaporfyrin (5 –COOH)	↑
U-Koproporfyrin (4 –COOH)	



Pyrrole







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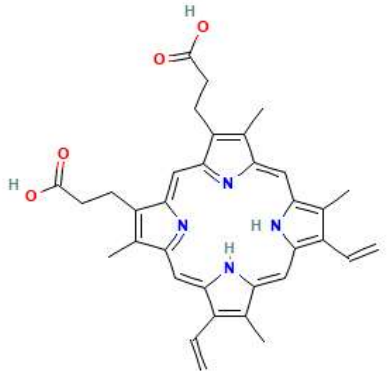
protoporphyrin IX

PubChem protoporphyrin IX (Compound)

1.1 2D Structure

Structure Search | Get Image

Chemical Structure Depiction



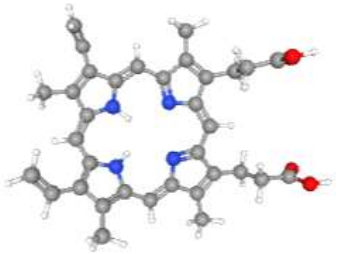
PubChem

1.2 3D Conformer

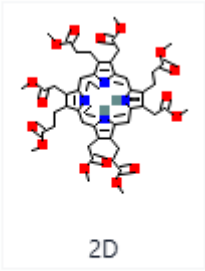
Structure Search | Get Image

Interactive Chemical Structure Model

- (●) Ball and Stick
- ( ) Sticks
- ( ) Wire-Frame
- ( ) Space-Filling
- [x] Show Hydrogens
- [ ] Animate

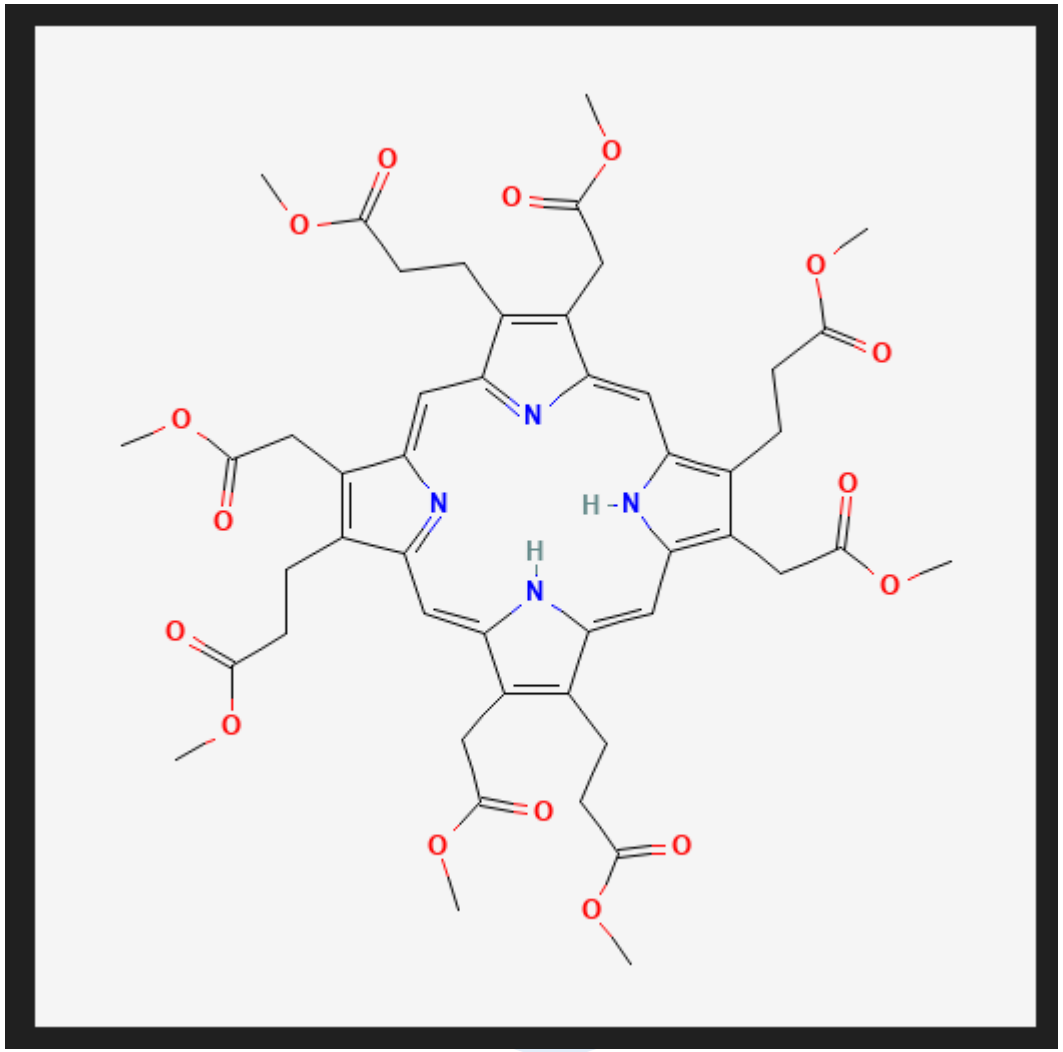


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2D

$C_{48}H_{54}N_4O_{16}$

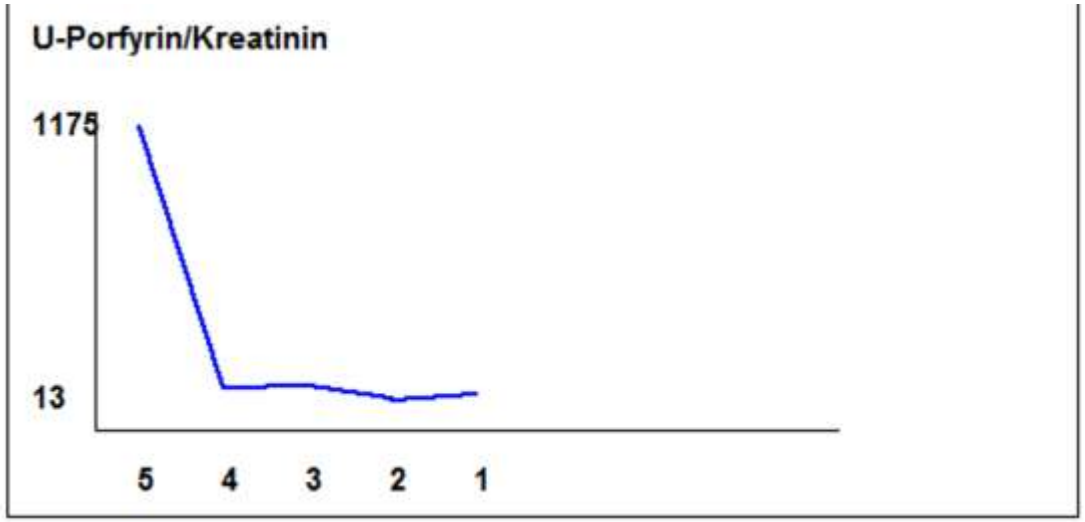


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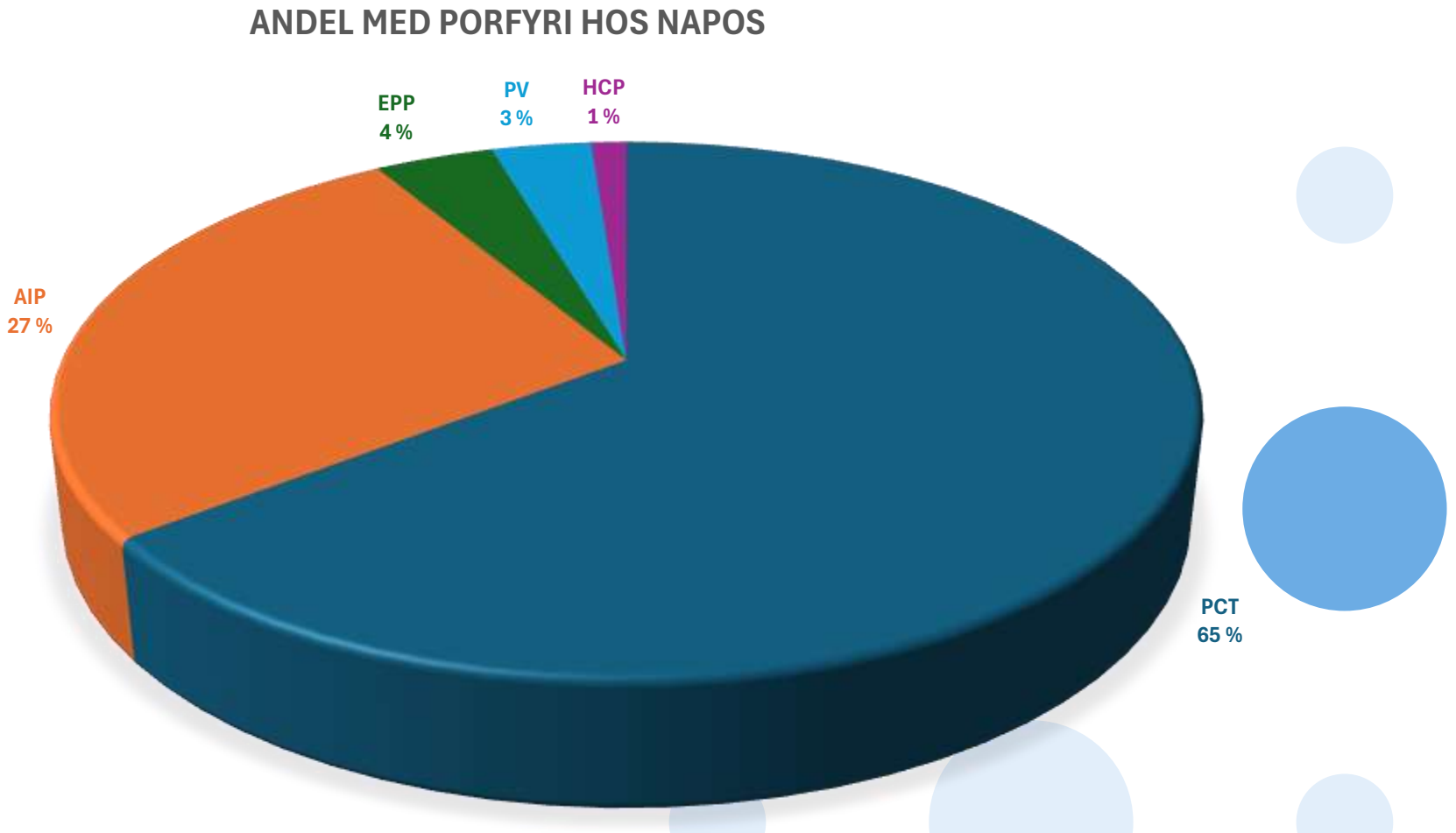
PubChem Uroporfyrrin

Under behandling (blodtapping/klorokin)

- Hudplagene forsvinner først
- Kan ta 1-1.5 år før mengden porfyriner i urin normaliseres

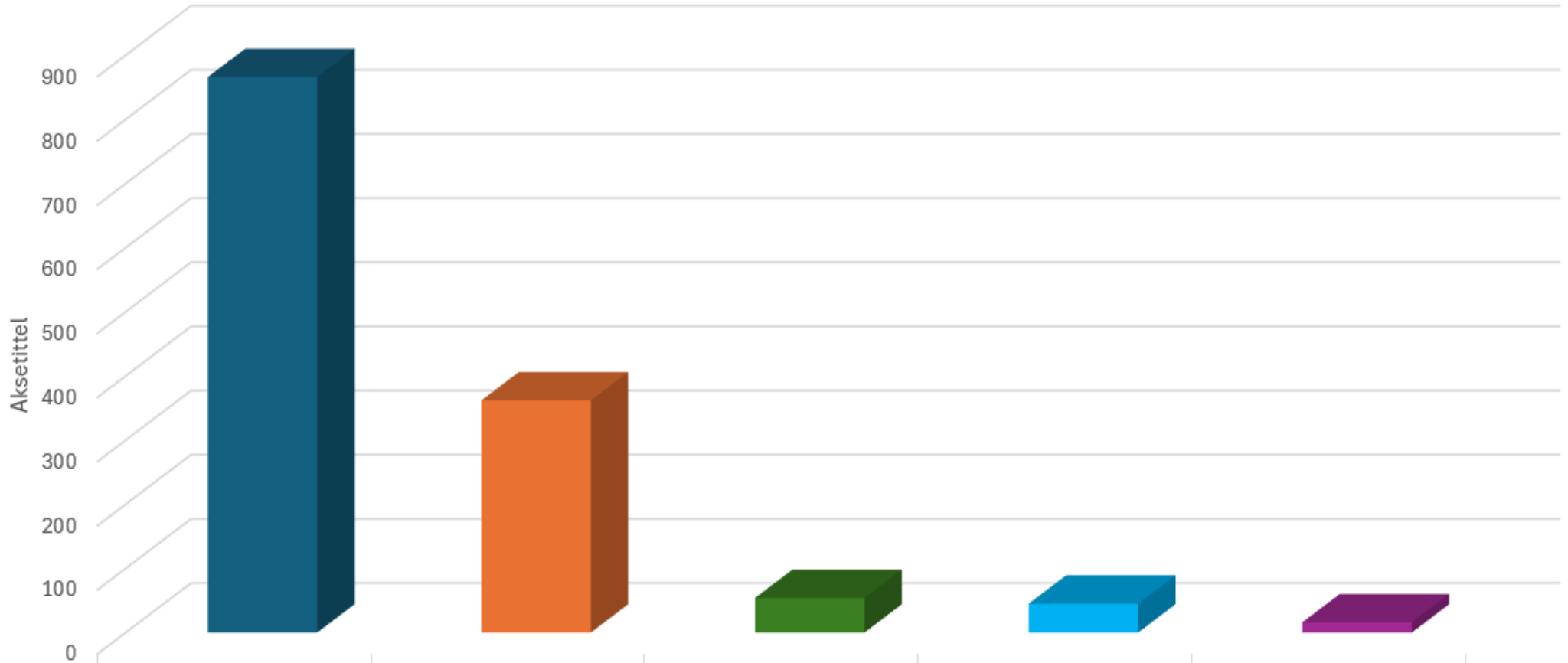


Antall

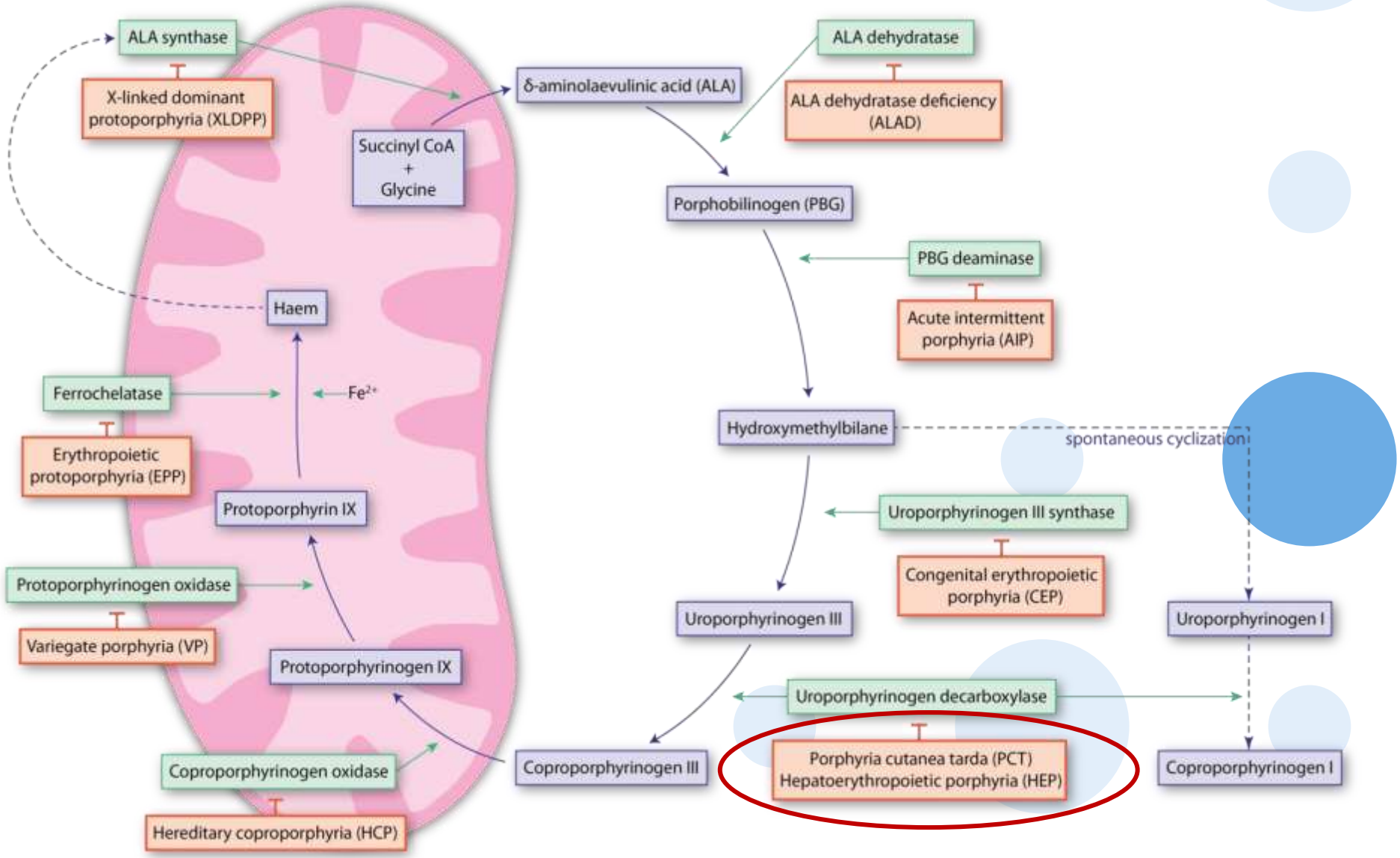


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Antall med porfyri hos NAPOS



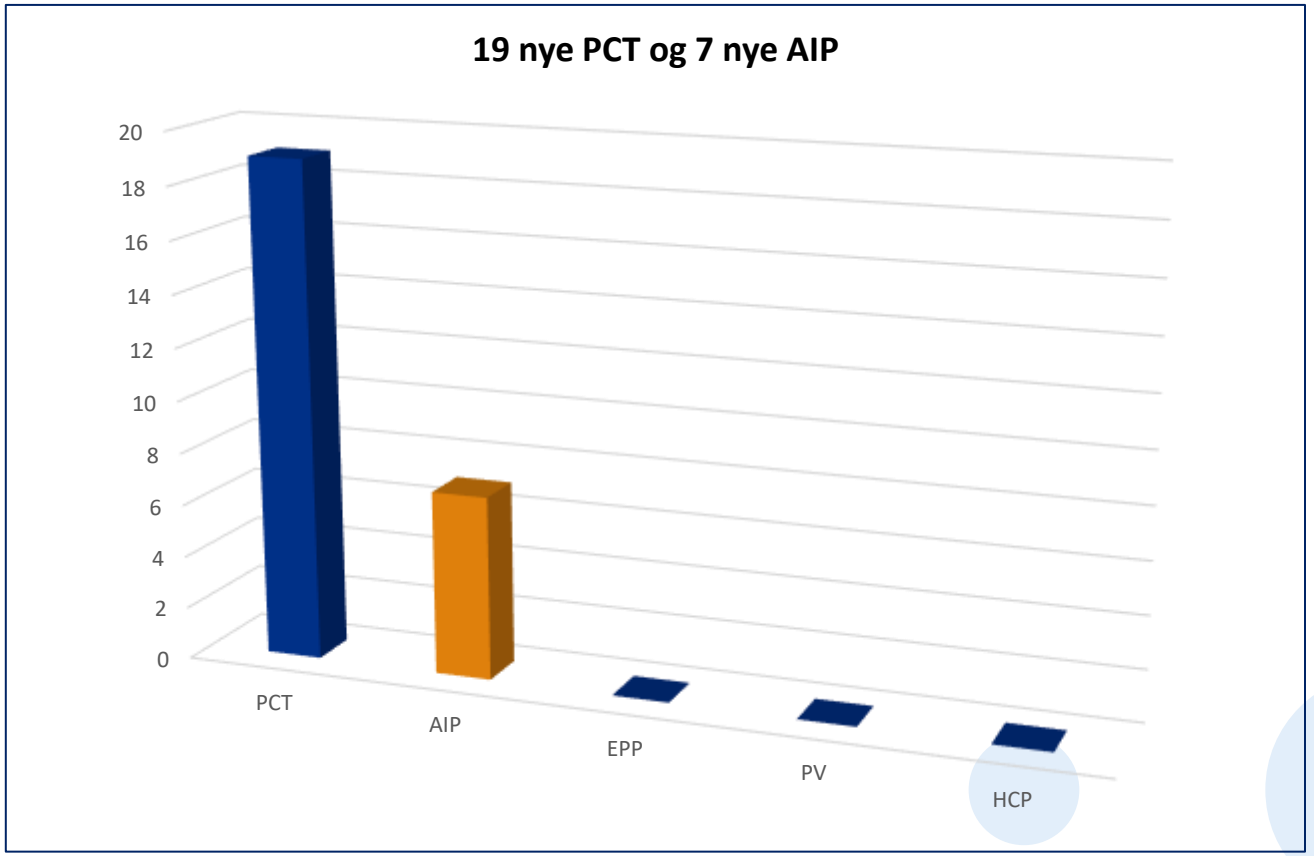
	PCT	AIP	EPP	PV	HCP
Antall med porfyri hos NAPOS	866	362	54	45	16



Helene Bustad Johannessen

NASJONALT SENTER FOR
SJELDNE DIAGNOSER

Hva fant vi i 2025 ?



NASJONALT SENTER FOR
SJELDNE DIAGNOSER

Laboratoriehåndboken Helse Stavanger

ferr

Analyser | Labnytt | Bestill prøvetakingsutstyr



Serumrør med gel 5 ml, gul kork

Ferritin

Relatert: (096)

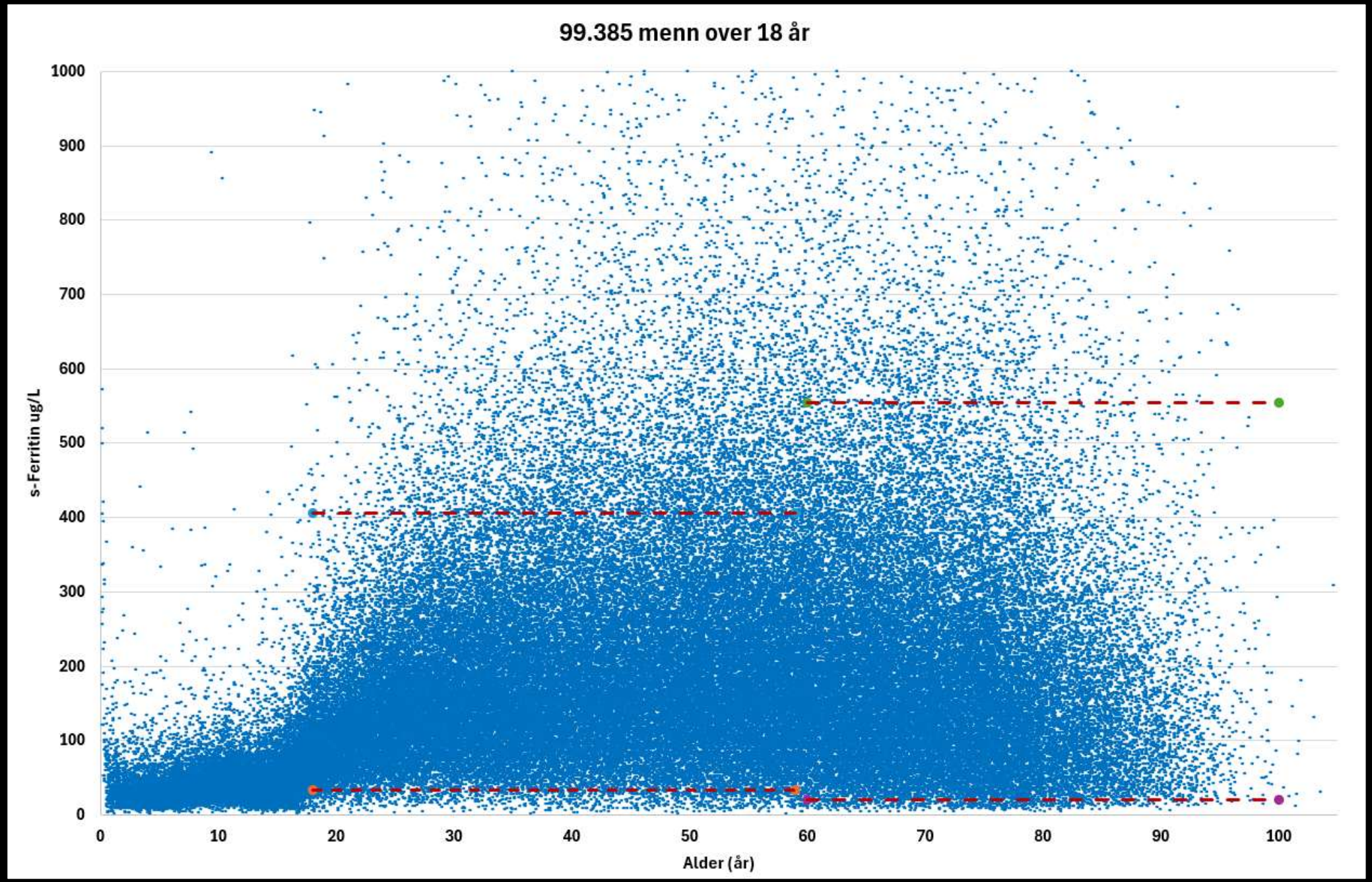
Prøvemateriale: Serum

Referanseområde:

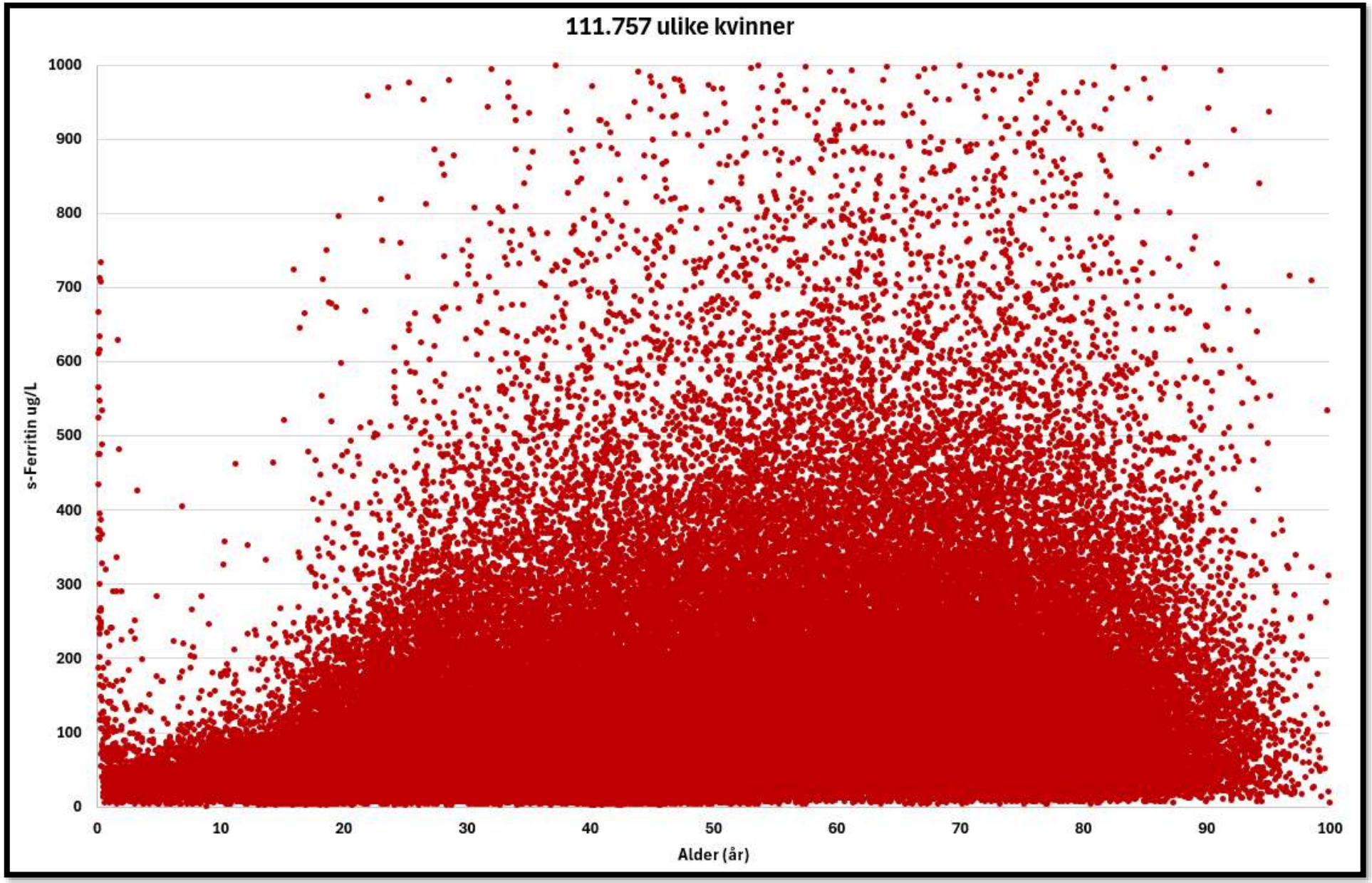
Kvinner: >= 19 - 52 år: 10 - 218 ug/L
Kvinner: >= 53 år: 23 - 383 ug/L
Menn: >=20 - 59 år: 33 - 406 ug/L
Menn: >= 60 år: 21 - 555 ug/L

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Stavanger universitetssjukehus

Ferritin

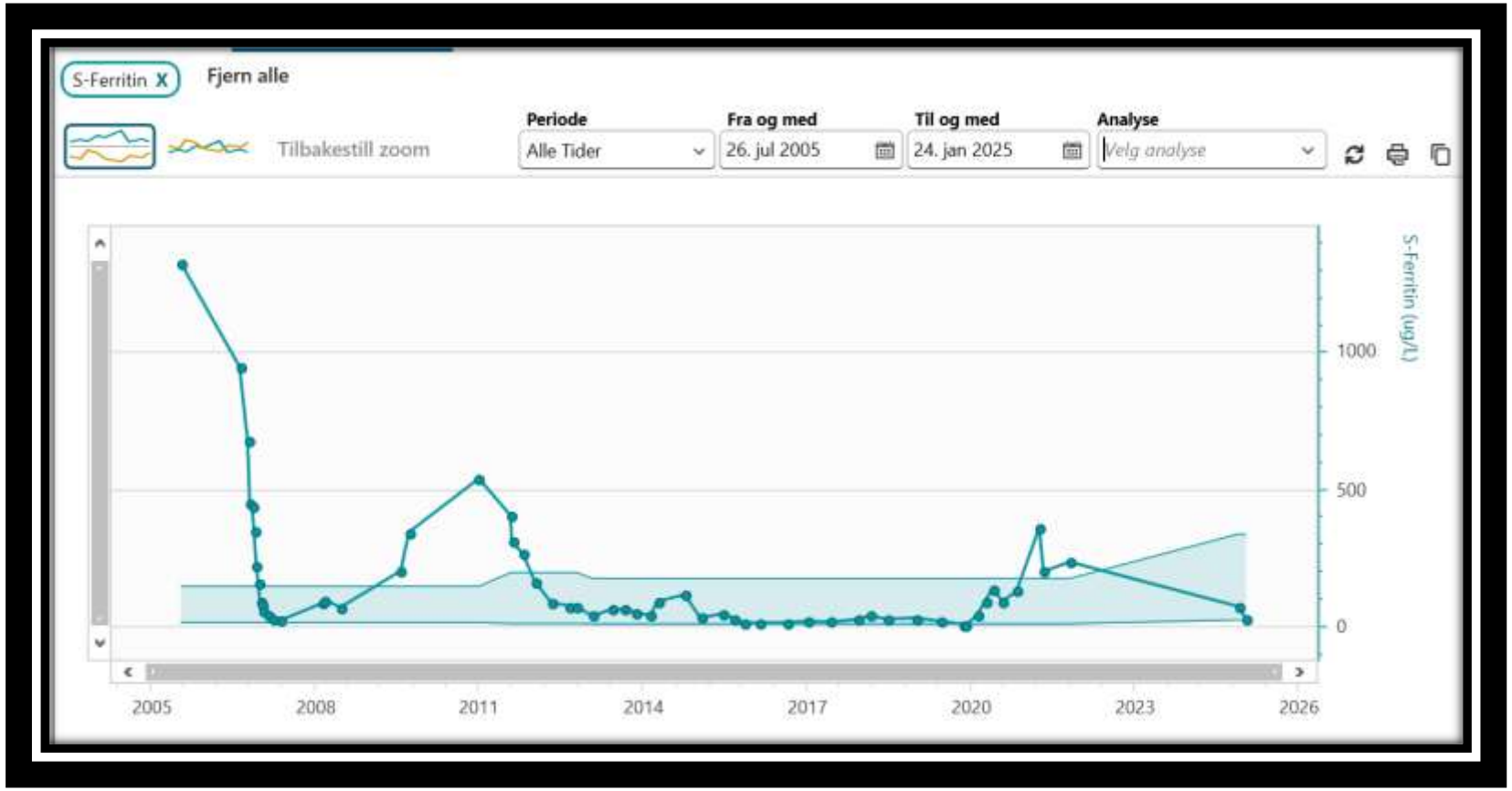

99.385 menn over 18 år
s-Ferritin ug/L
1000
900
800
700
600
500
400
300
200
100
0
0
10
20
30
40
50
60
70
80
90
100
Alder (år)

Kvinner



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Viktig å gå til kontroll og passe på at ferritin ikke stiger igjen



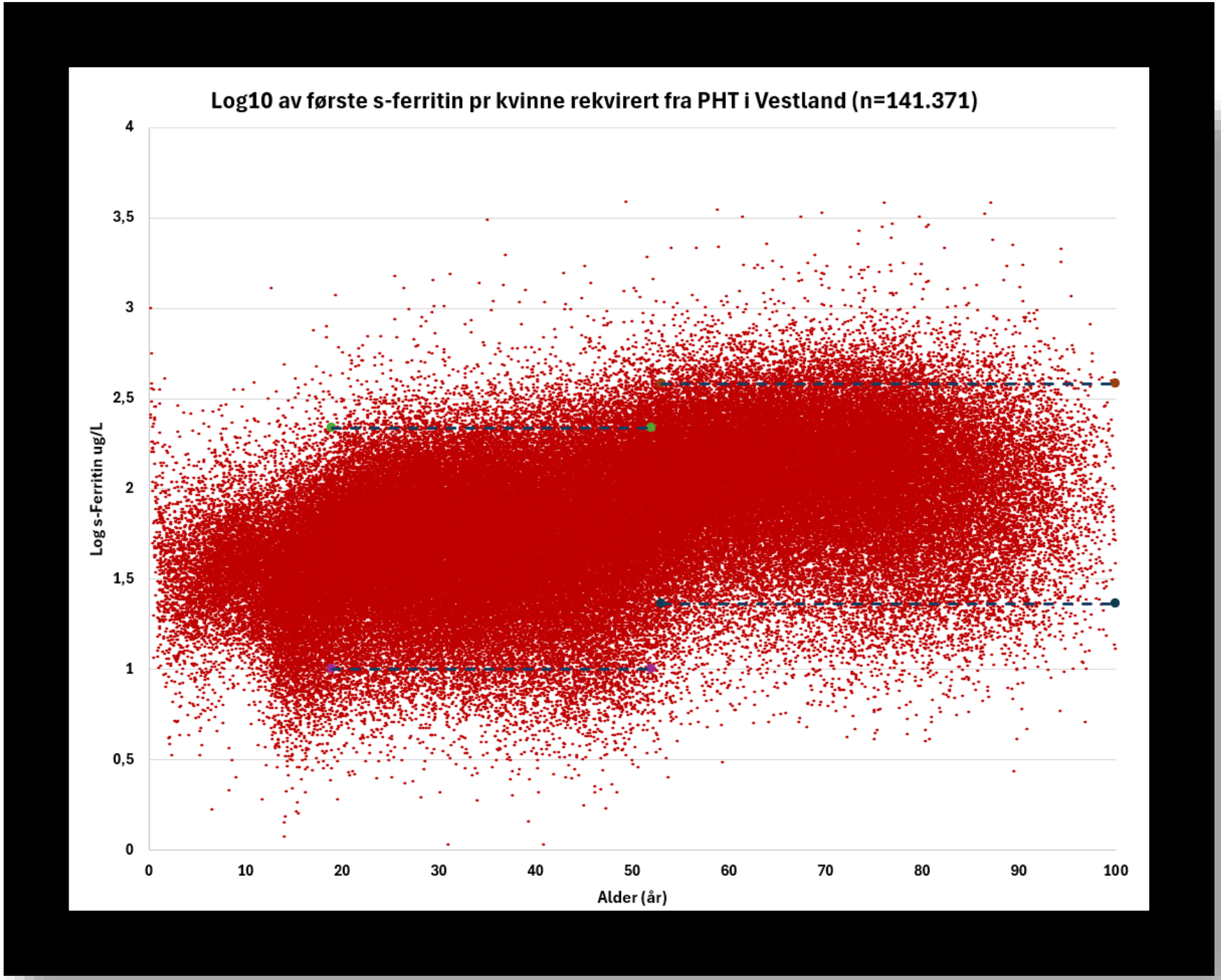
HELSE STAVANGER
Stavanger universitetssjukehus

Ferritin stiger forbigående ved sykdom

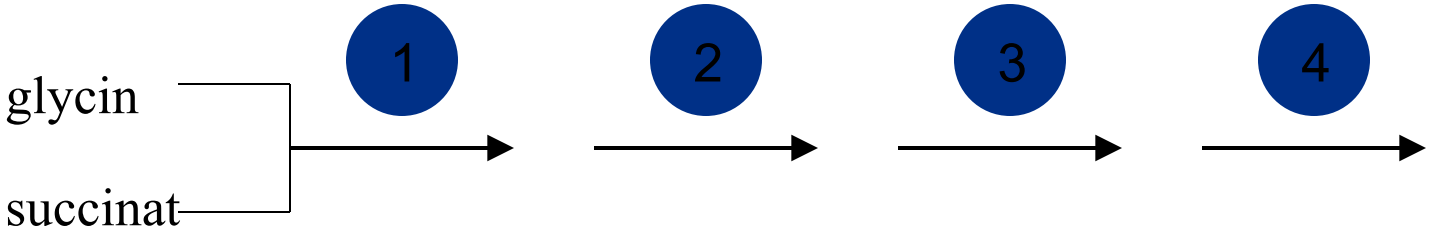
Dette gir ikke fare for hudsymptomer

Ferritin faller igjen når man blir frisk

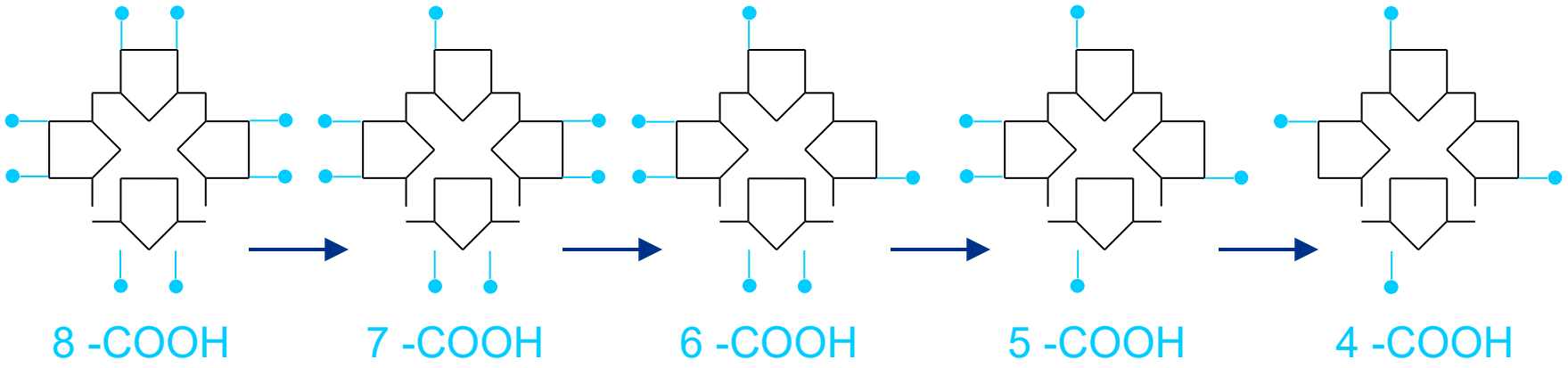
Logaritmen av Ferritin



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5 *Uroporfyrinogen dekarboksylase (Uro-D)*





PCT er mest en leversykdom som gir symptomer fra huden

Ofte flere årsaker til at PCT blir utløst:

- Arv med redusert 5. enzym fra mor eller far
- For høyt lager av jern i leveren
- For stor alkohol inntak
- Østrogener (p-piller)
- Virus sykdommer