

POLYMORPHISMS OF ALPHA-1-ANTITRYPSIN AND TRANSFERRIN IN
NORTH-WEST GERMANY

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Aims of the study:

- Investigation of the applicability of Immobiline dry plates for the determination of alpha-1-antitrypsin(Pi) and transferrin(Tf) polymorphisms.
The system to define alpha-1-antitrypsin gene products was also set up in view of the diagnosis and prevention of premature emphysema and progressive liver disease.
- Population genetics of alpha-1-antitrypsin and transferrin in North-West Germany.

Alpha-1-Antitrypsin

Method: Electrofocusing in Immobiline gel, pH-range 4.2-4.9 (LKB, Bromma, Sweden).

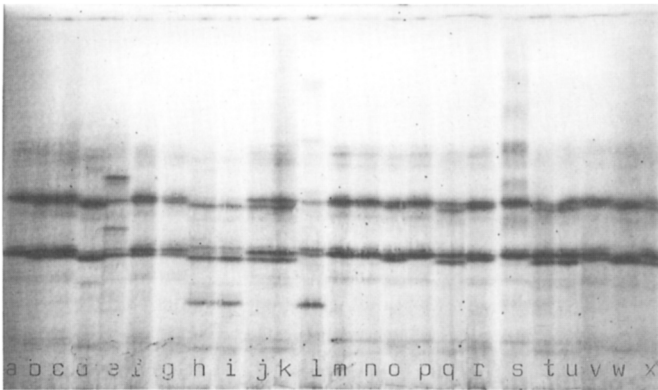
1. Rehydration: Immobiline dry plate 16 hours in the reswelling cassette with 25% glycerol.
2. Sample application: 5 µl serum were applied to filter paper pieces, laid on the gel surface about 1 cm from the cathode, by using a multiple microlitre syringe (6x50 µl Hamilton).
3. Electrode solutions: Anode and cathode: distilled water
4. Running conditions: Electrical settings: 5000 V, 2 mA, 5 W
Cooling plate: 8° C
Running time: 1 hour with sample application pieces,
6 hours without.
No prefocusing

Transferrin

Method: Electrofocusing in Immobiline gel, pH-range 5.0-6.0 (LKB, Bromma, Sweden)

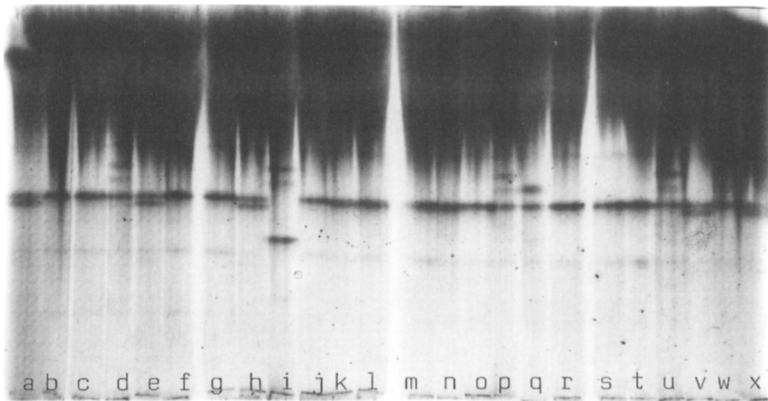
1. Sample preparation: 5 µl serum and 25 µl 0.25% ferric ammonium sulphate over night at 4°C.
2. Rehydration: Immobiline dry plate 2 hours in the reswelling cassette with 25% glycerol.
3. Sample application: 5 µl of the prepared serum were applied to filter paper pieces, laid on the gel surface about 1 cm from the cathode, by using a multiple microlitre syringe (6x50 µl Hamilton)
4. Electrode solutions: Anode and cathode: distilled water
5. Running conditions: Electrical settings: 3000 V, 4 mA, 12 W
Cooling plate: 8° C
Running time: 1 hour with sample application pieces,
3.5 hours without.
No prefocusing

Fixation, staining, destaining and preservation according to LKB-Instructions(1804)



Alpha-1-Antitrypsin phenotypes in figure 1:

a:M1-1,b:M3-1,c:M3-1,d:M2-2,e:M3-F,f:M3-1,g:M3-1,h:M2-S,i:M2-S,
j:M2-1,k:M2-1,l:M1-S,m:M1-1,n:M1-1,o:M3-1,p:M1-1,q:M2-1,r:M3-1,
s:M1-1,t:M2-1,u:M2-1,v:M3-1,w:M3-2,x:M2-1.



Transferrin phenotypes in figure 2:

a:C2-1,b:C1-1,c:C1-1,d:C1-B,e:C2-1,f:C1-1,g:C1-1,h:C2-1,i:B-D,
j:C1-1,k:C3-1,l:C3-1,m:C3-1,n:C3-1,o:C1-1,p:C1-B,q:C1-B,r:C1-1,
s:C1-1,t:C1-1,u:C1-B,v:C2-1,w:C1-1,x:C2-1

Results:

For alpha-1-antitrypsin the gene products M1, M2, M3, F, S and Z could be defined.

Typing a group of 500 healthy,unrelated blood donors 13 different phenotypes(M1-1,M2-1,M3-1,M1-F,M1-S,M1-Z,M2-2,M2-F, M2-S,M3-2,M3-3,M3-F,M3-S)were found.

The observed values agreed well with the expected ones,assuming

