'A provisional exclusion map - the by-product of attempts at deletion mapping' by Professor Malcolm Andrew Ferguson-Smith

Publication/Creation

c.1960s-c.1970s

Persistent URL

https://wellcomecollection.org/works/uzcpbdcd

License and attribution

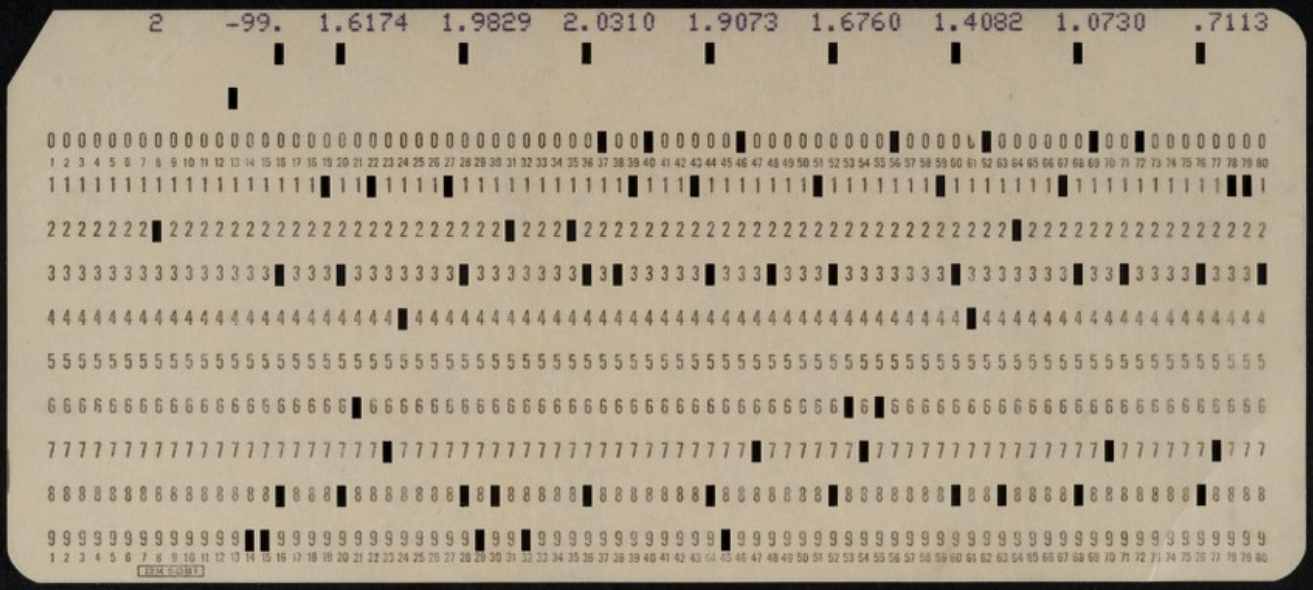
You have permission to make copies of this work under a Creative Commons, Attribution, Non-commercial license.

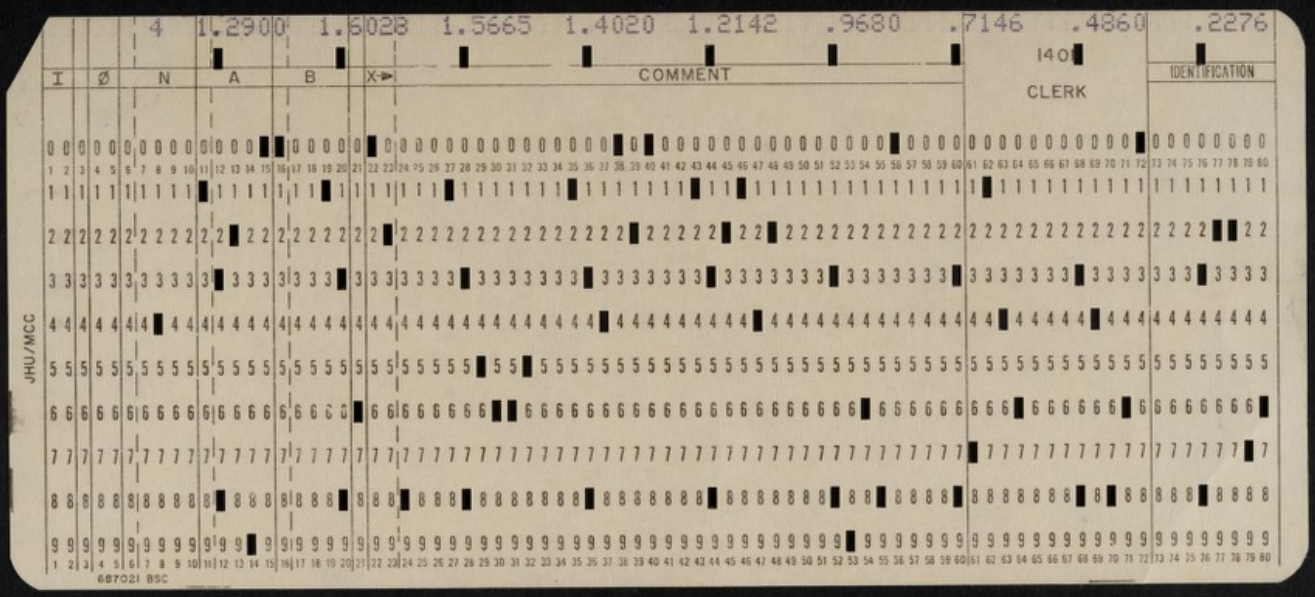
Non-commercial use includes private study, academic research, teaching, and other activities that are not primarily intended for, or directed towards, commercial advantage or private monetary compensation. See the Legal Code for further information.

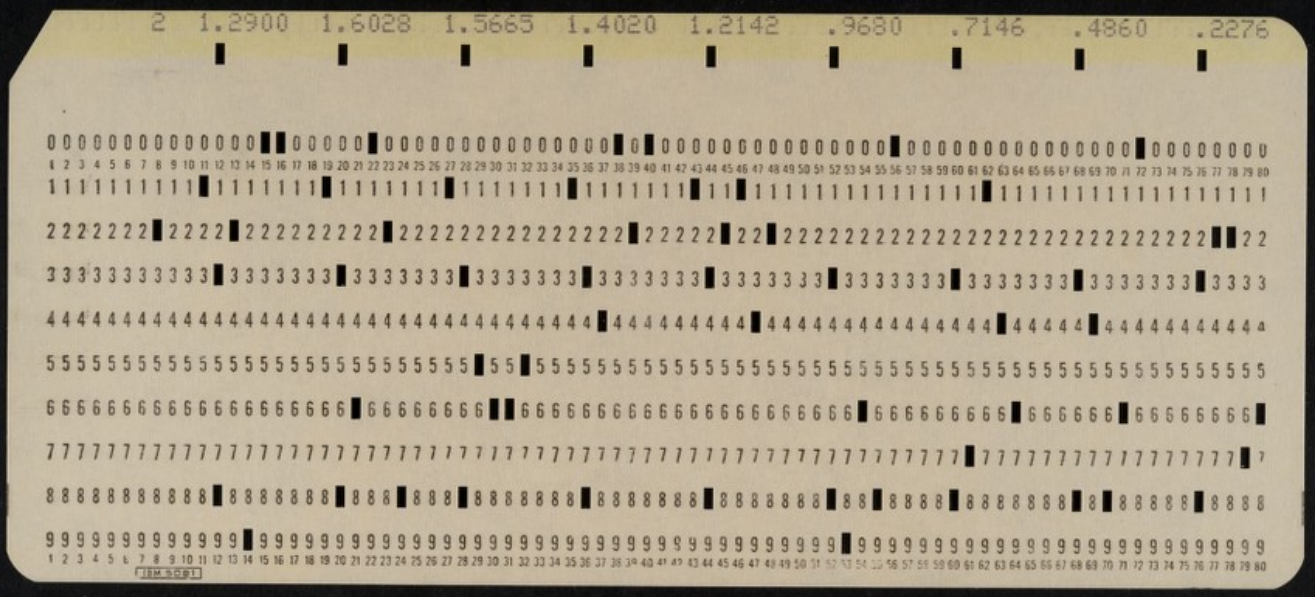
Image source should be attributed as specified in the full catalogue record. If no source is given the image should be attributed to Wellcome Collection.

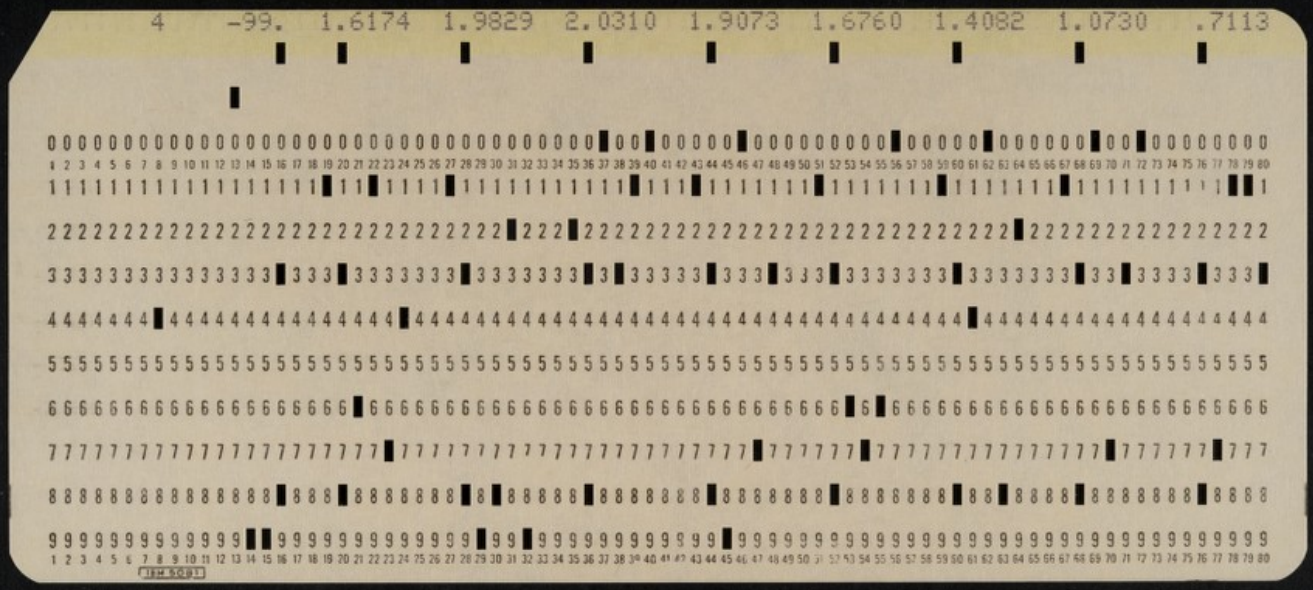
wellcome collection

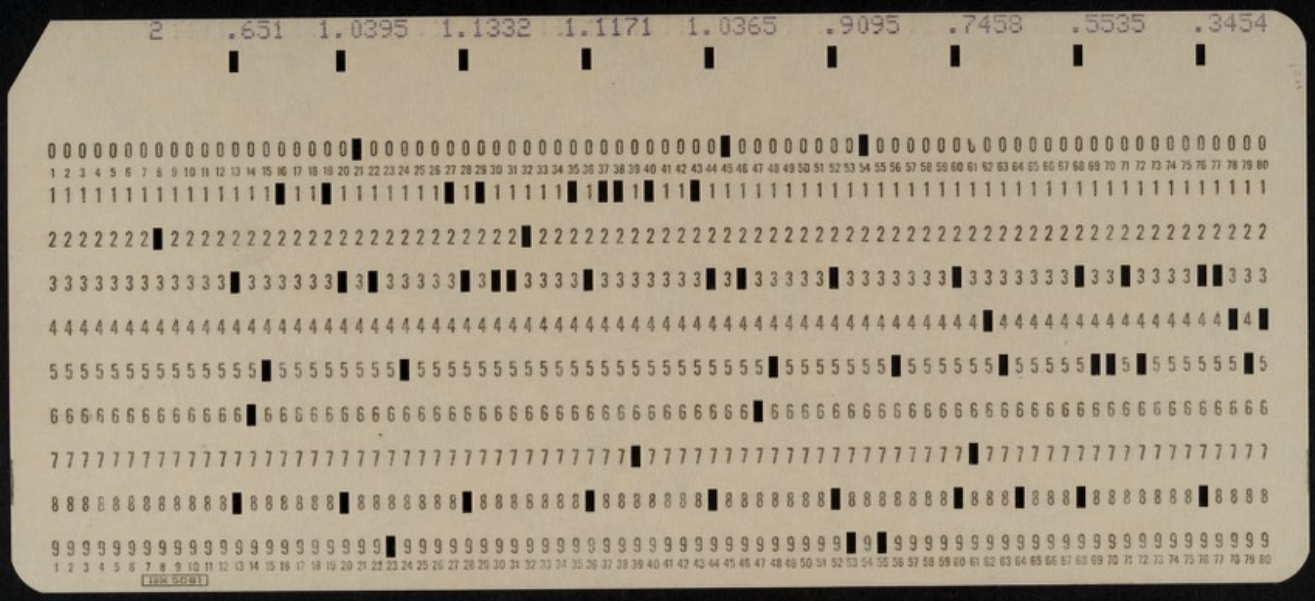
Wellcome Collection
183 Euston Road
London NW1 2BE UK
T +44 (0)20 7611 8722
E library@wellcomecollection.org
https://wellcomecollection.org

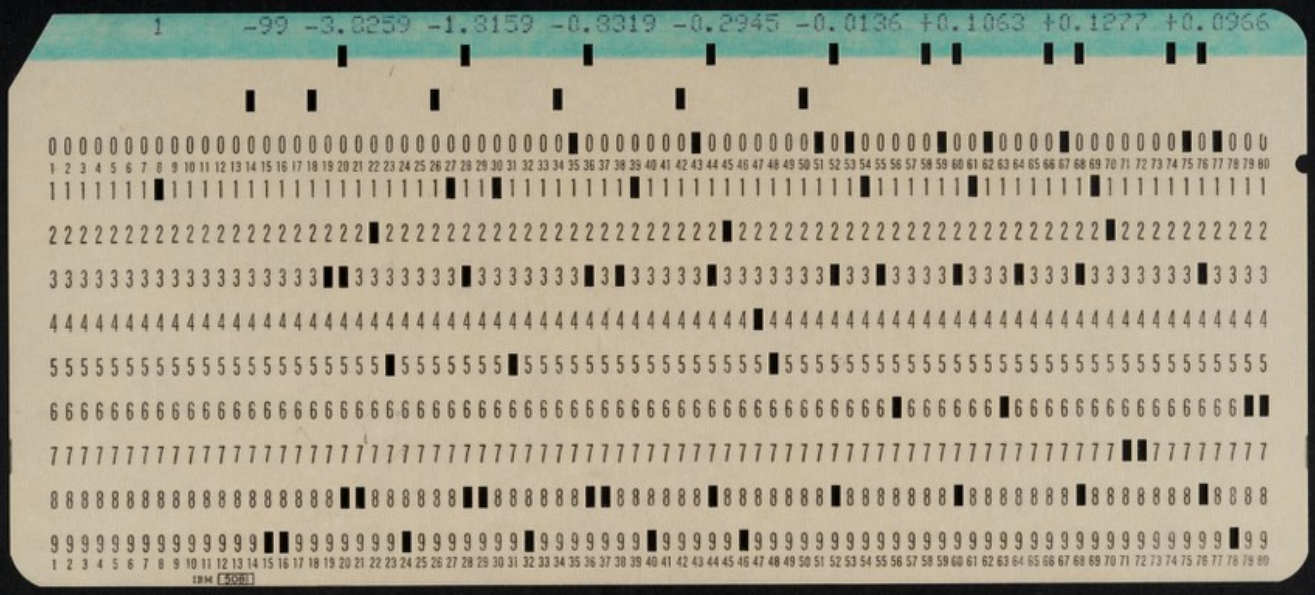

2 -99. 1.6174 1.9829 2.0310 1.9073 1.6760 1.4082 1.0730 .7113


4 1.2900 1.6028 1.5665 1.4020 1.2142 .9680 .7146 .4860 .2276
I
Ø
N
A
B
X
COMMENT
1401
CLERK
IDENTIFICATION
JHU/MCC
627021 BSC

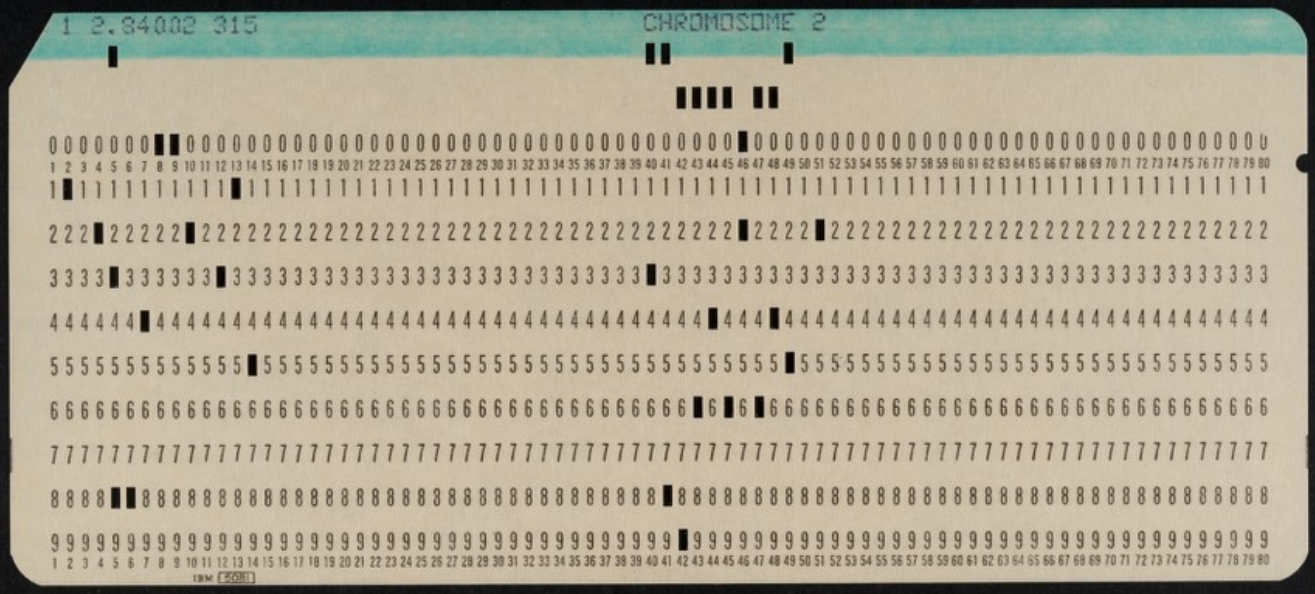

2 1.2900 1.6028 1.5665 1.4020 1.2142 .9680 .7146 .4860 .2276

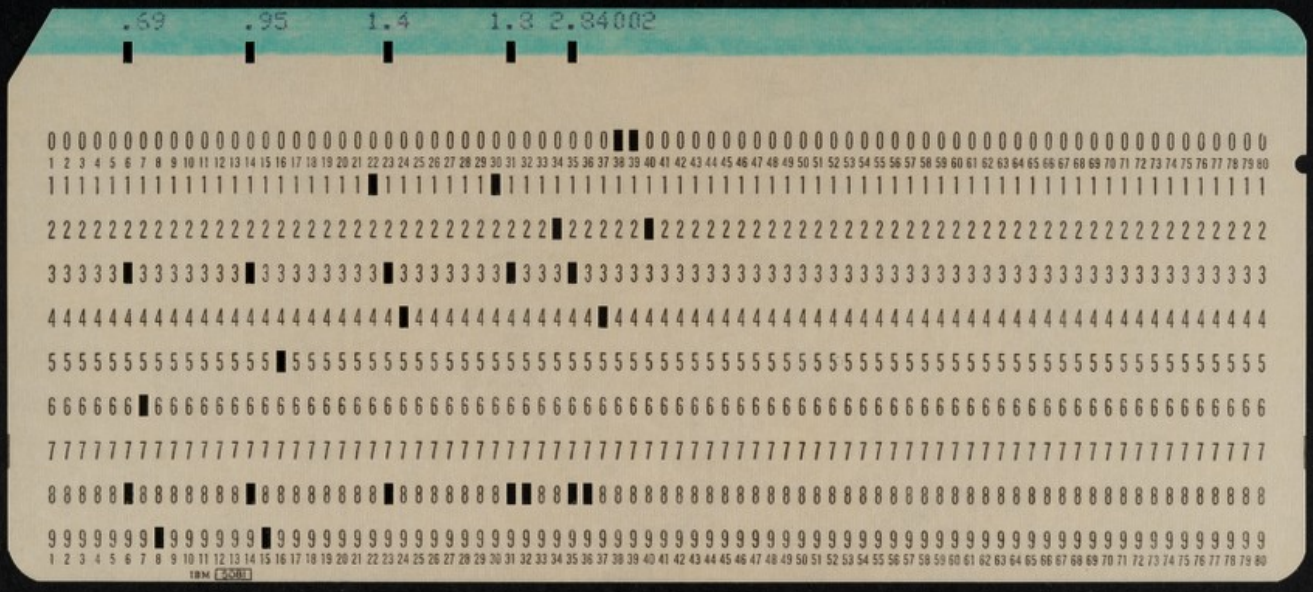

4 -99. 1.6174 1.9829 2.0310 1.9073 1.6760 1.4082 1.0730 .7113

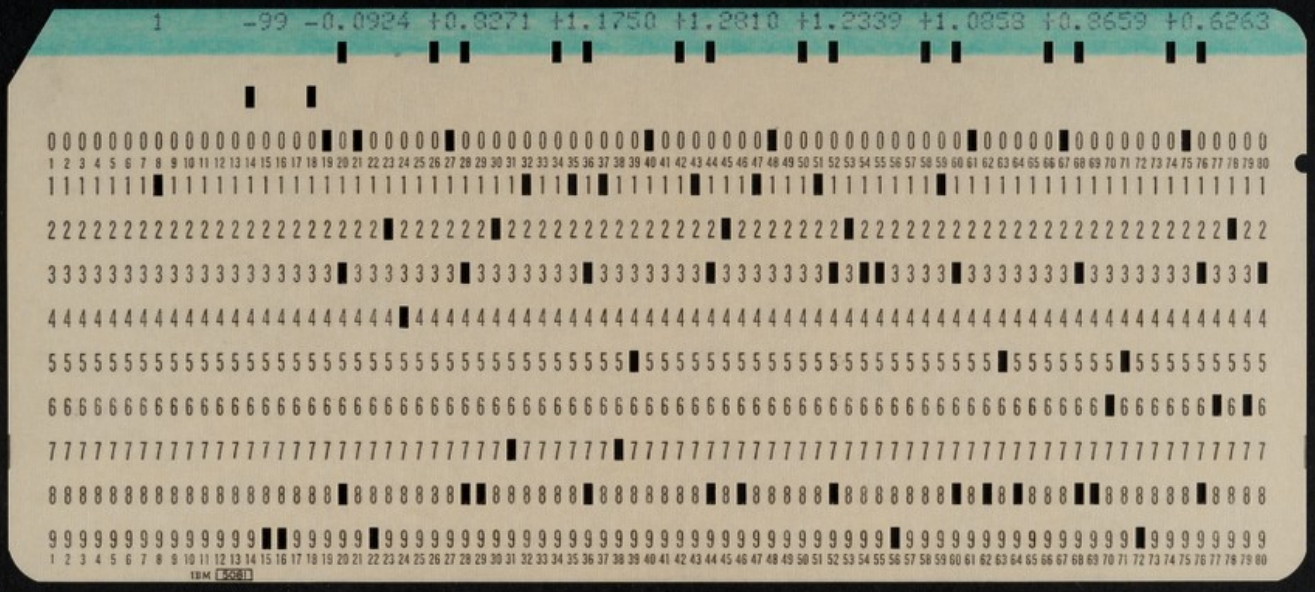

2 .651 1.0395 1.1332 1.1171 1.0365 .9095 .7458 .5535 .3454


1 -99 -3.8259 -1.8159 -0.8319 -0.2945 -0.0136 +0.1063 +0.1277 +0.0966
IBM 5081

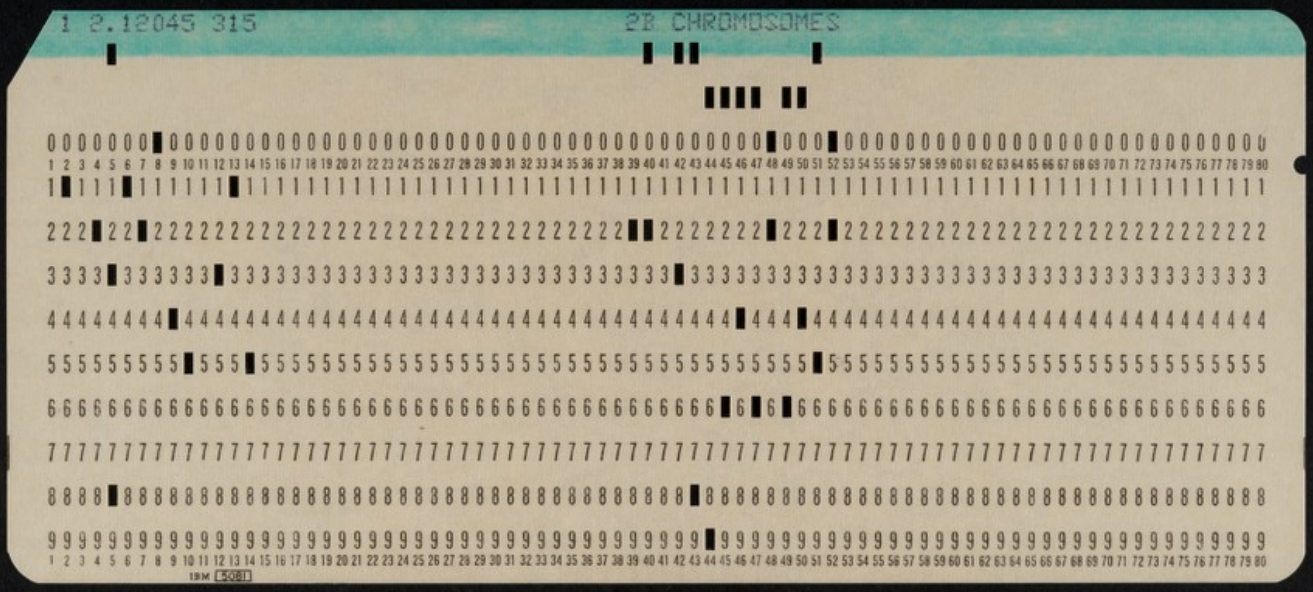
+0.0416 +0.0136 +0.0017
1/054
IBM 5081

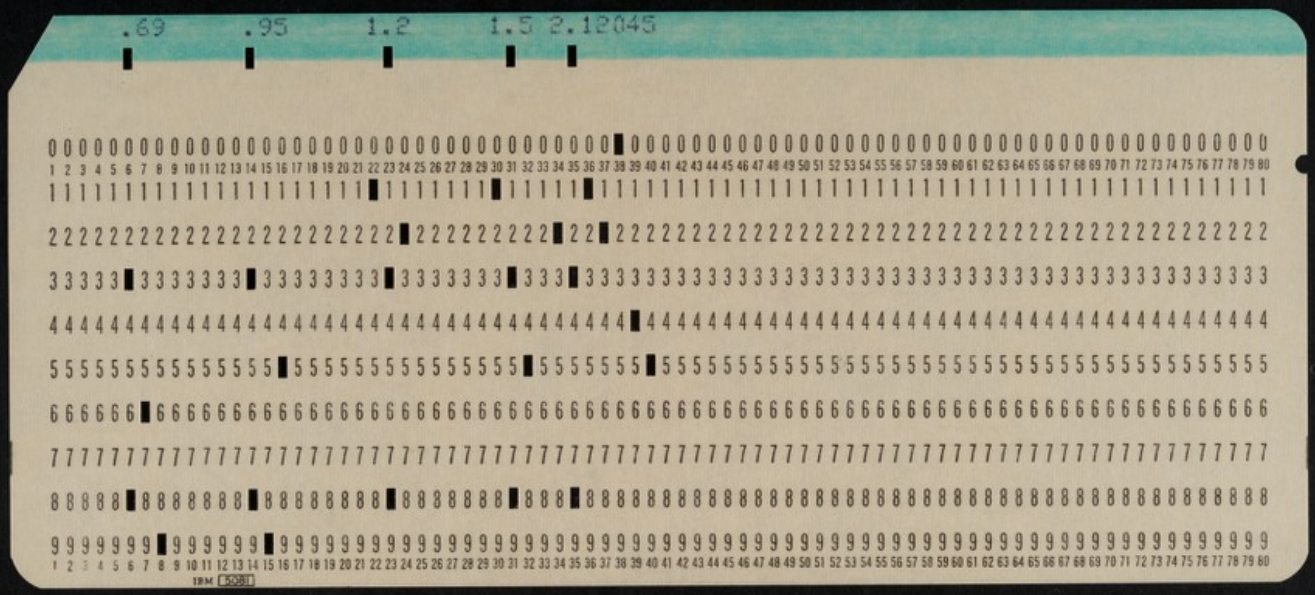

1 2.84002 315
CHROMOSOME 2

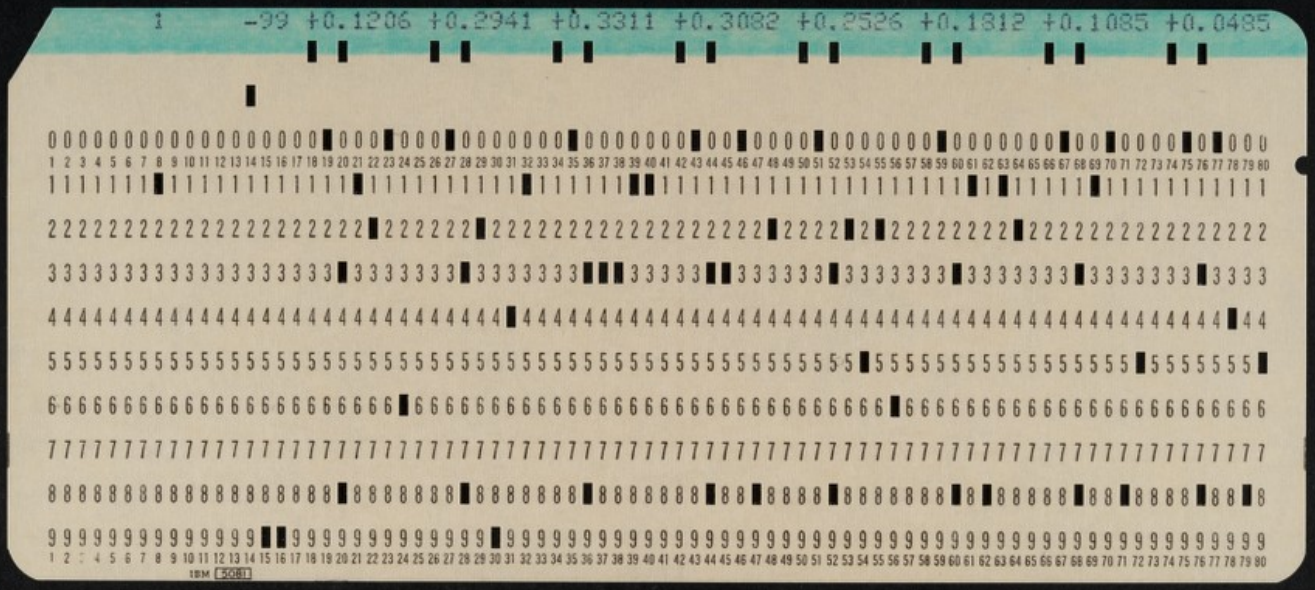

.69
.95
1.4
1.8
2.84002
IBM 3501

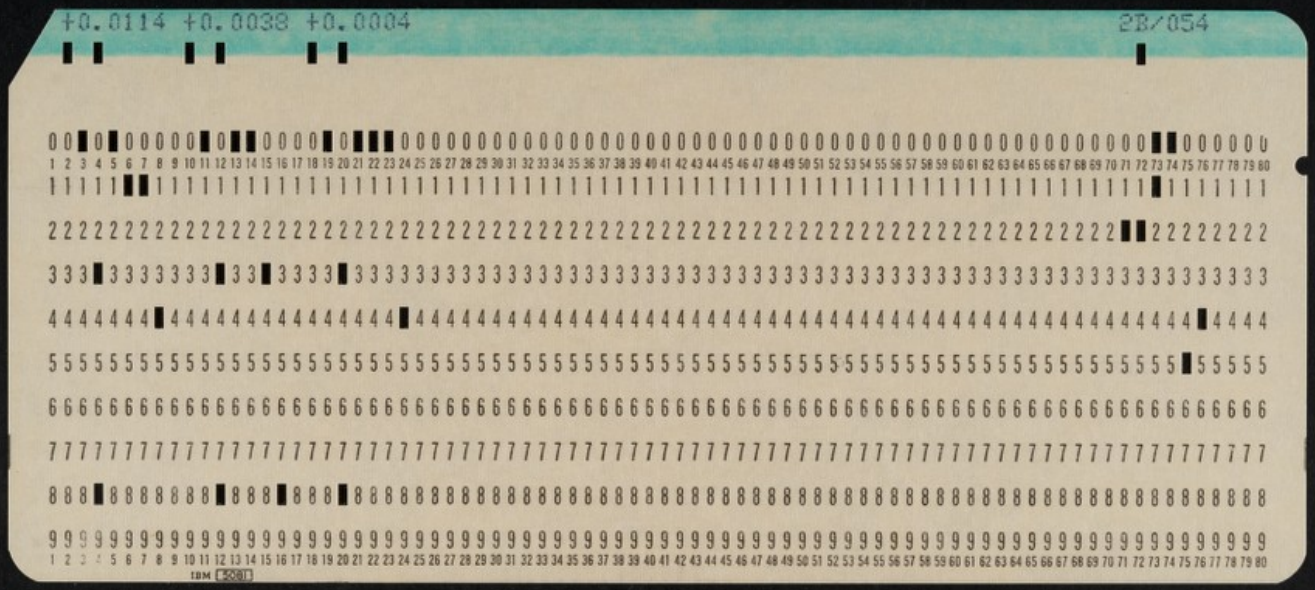

1 -99 -0.0924 +0.8271 +1.1750 +1.2810 +1.2339 +1.0858 +0.8659 +0.6263
IBM 3081

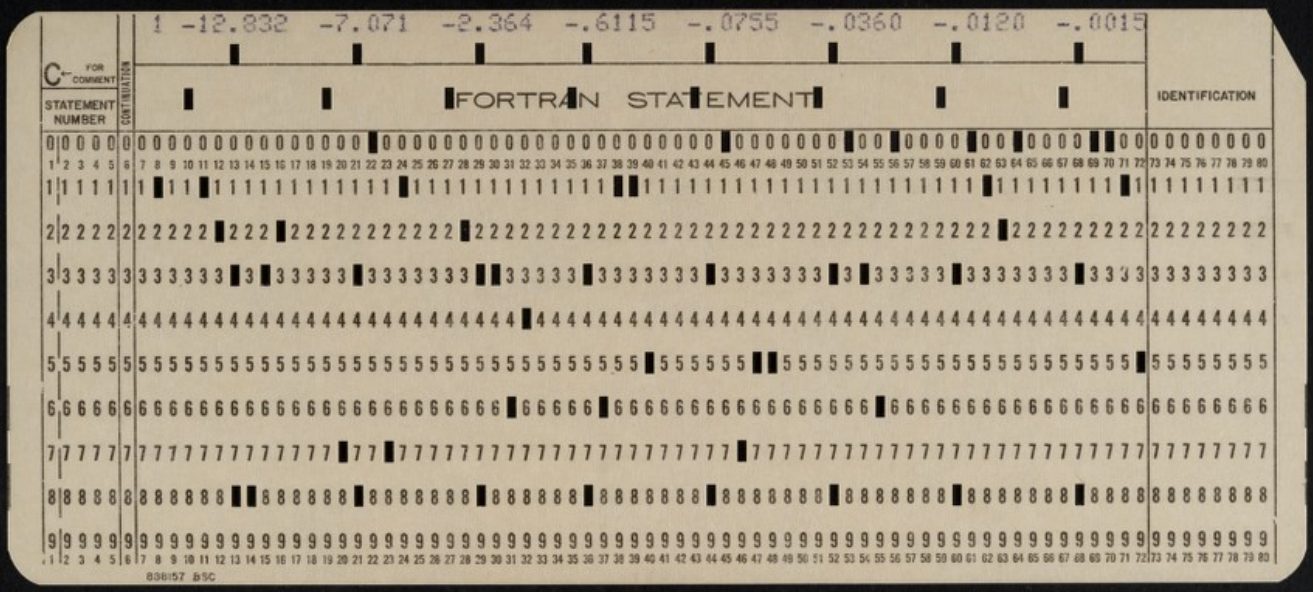
+0.3029 +0.0984 +0.0123
2/054
IBM (5081)

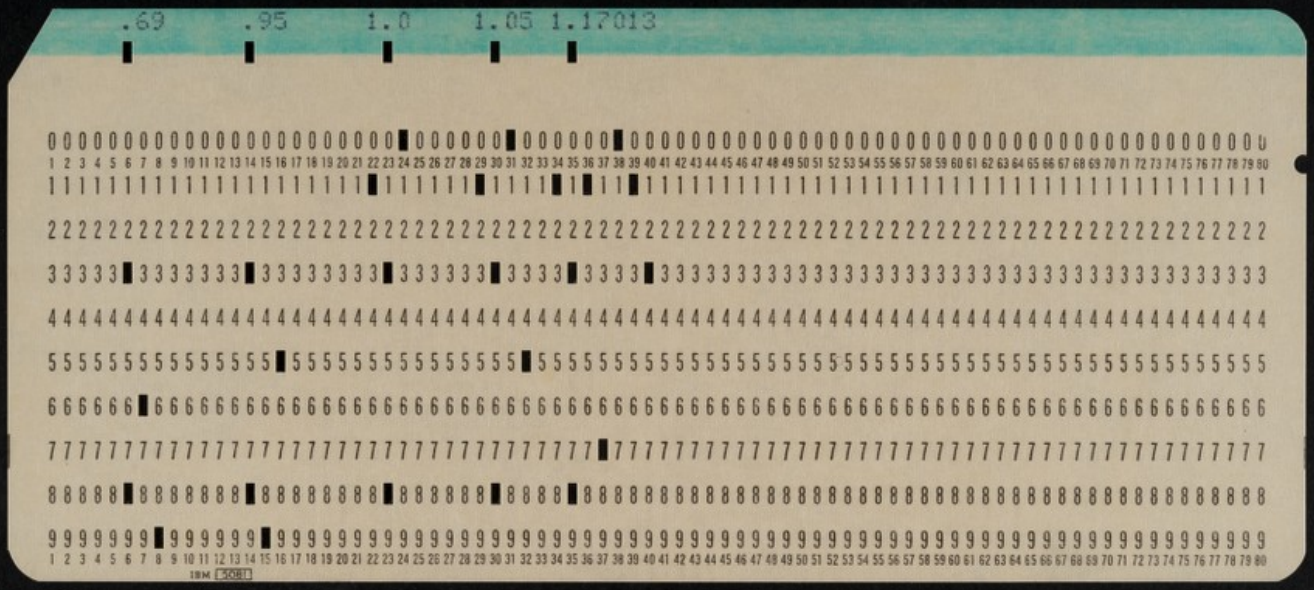

1 2.12045 315
2B CHROMOSOMES
IBM 5081

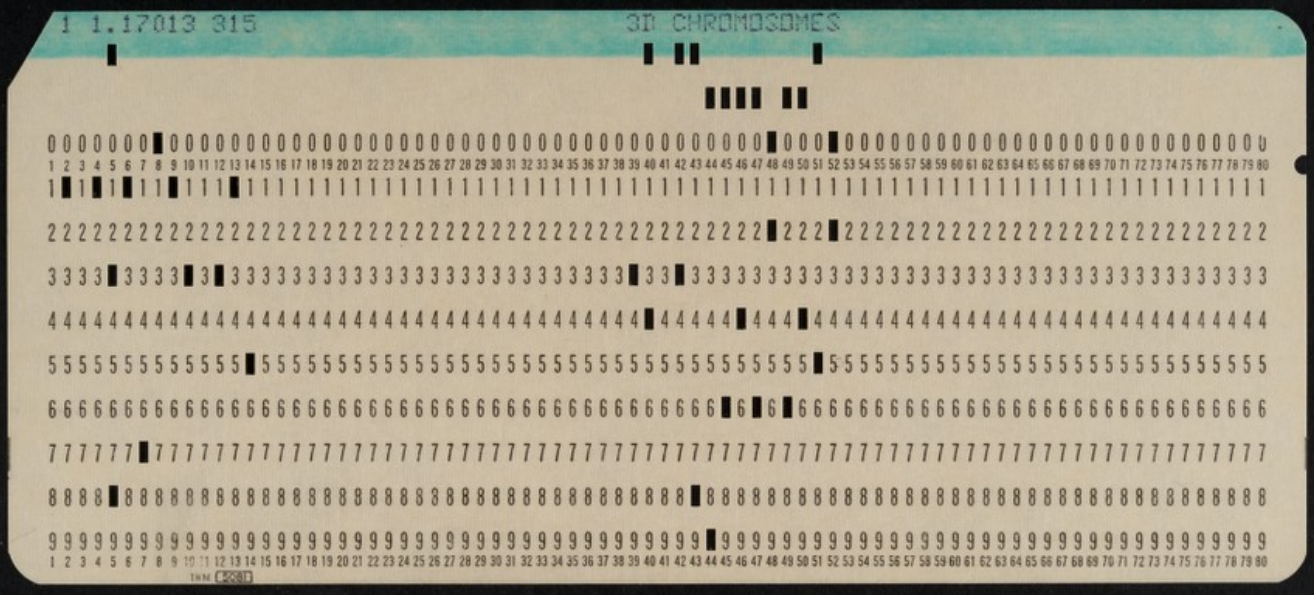

.69 .95 1.2 1.5 2.12045
IBM 5081

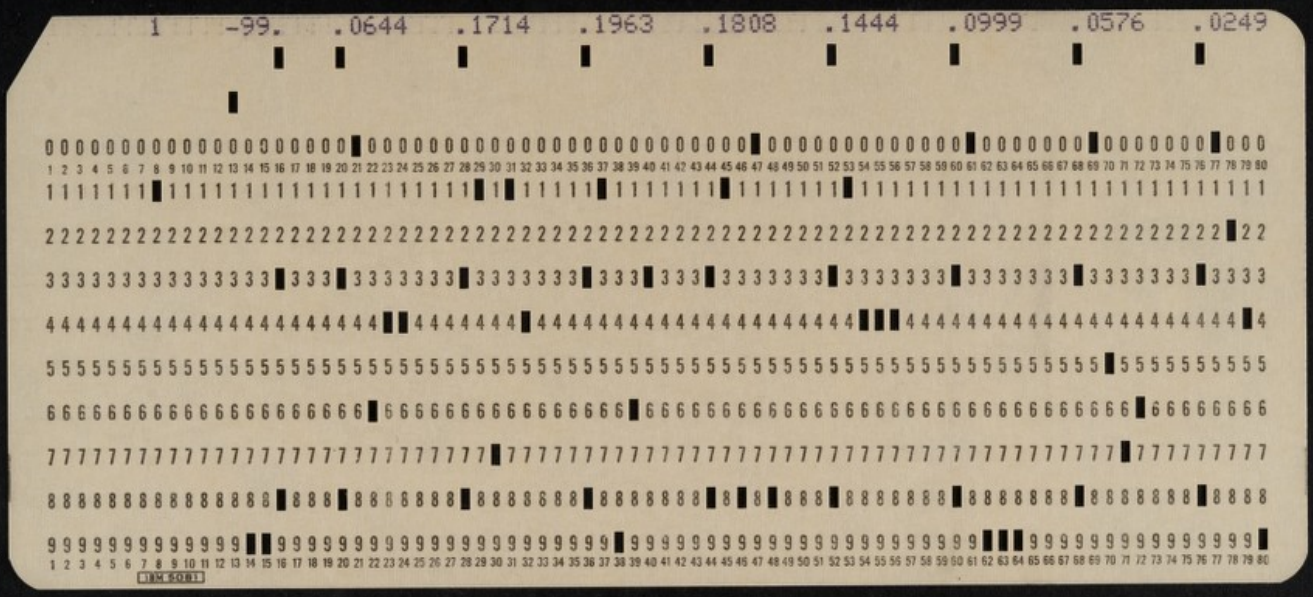

1 -99 +0.1206 +0.2941 +0.3311 +0.3082 +0.2526 +0.1812 +0.1085 +0.0485
IBM 5081

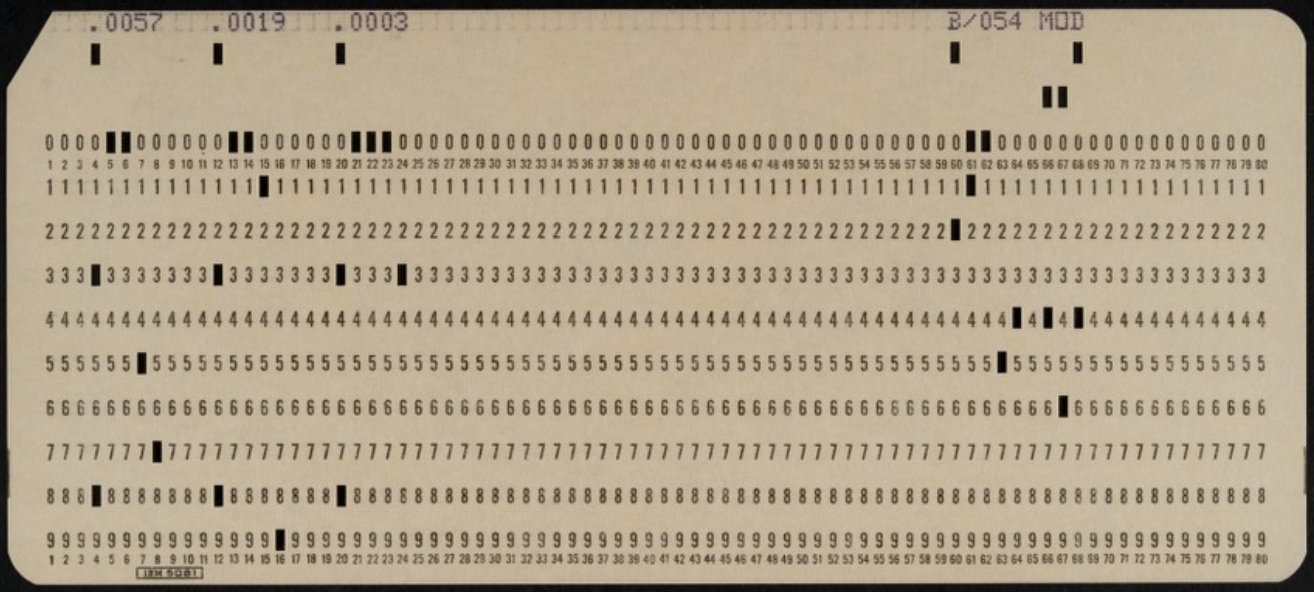

+0.0114 +0.0038 +0.0004
2B/054
IBM 5081

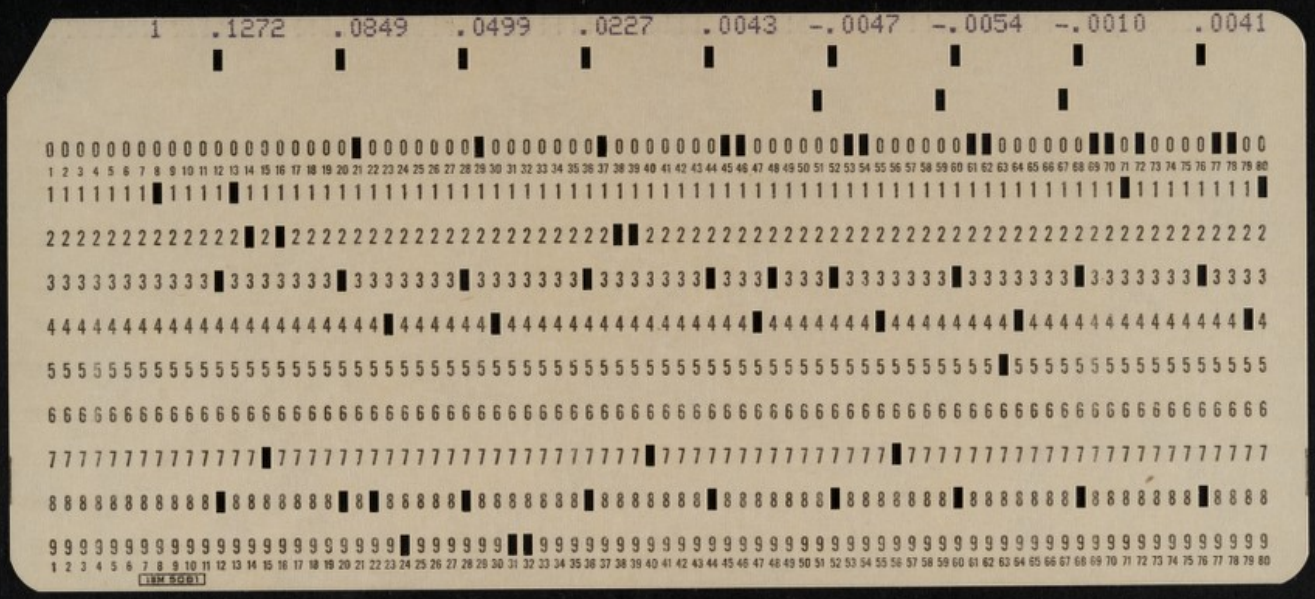

1 -12.832 -7.071 -2.364 -.6115 -.0755 -.0360 -.0120 -.0015
C FOR COMMENT
STATEMENT NUMBER
CONTINUATION
FORTRAN STATEMENT
IDENTIFICATION
888157 BSC

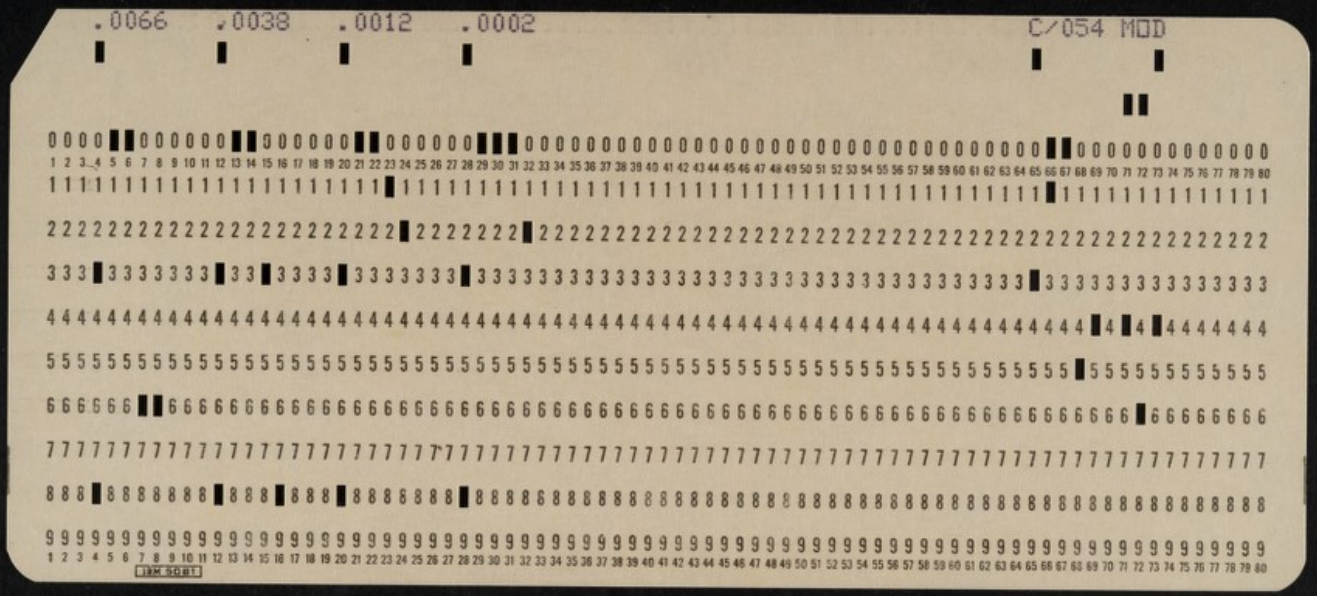

.69
.95
1.0
1.05
1.17013
IBM 3081

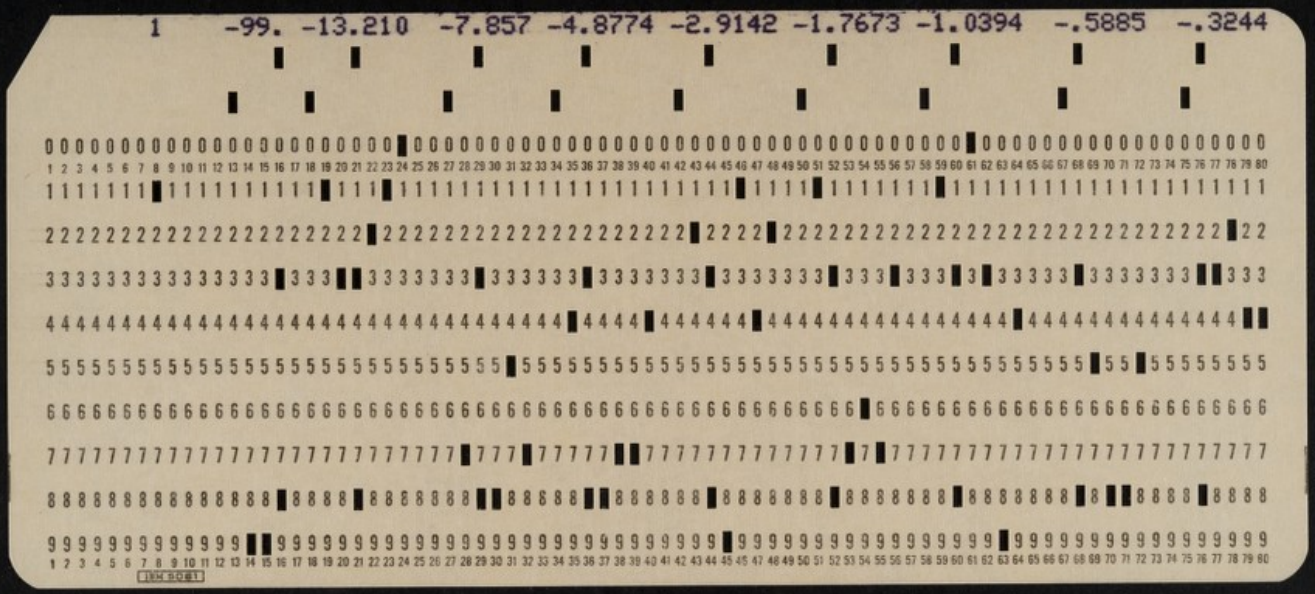

1 1.17013 315
3D CHROMOSOMES

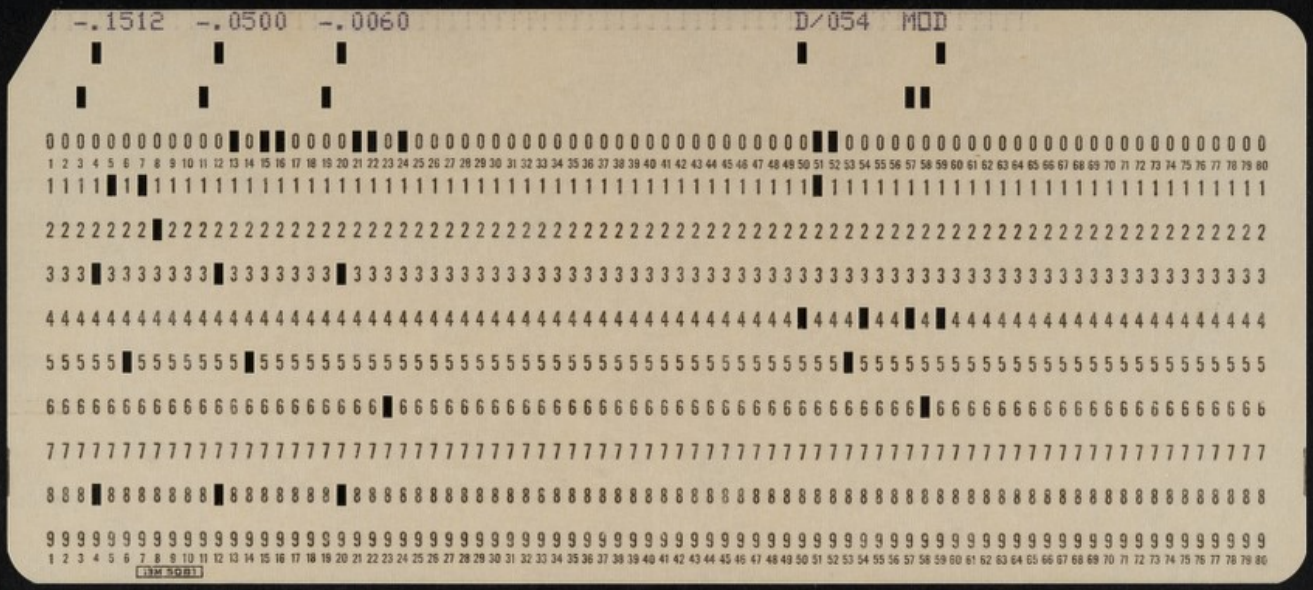

1 -99. .0644 .1714 .1963 .1808 .1444 .0999 .0576 .0249
IBM 5081

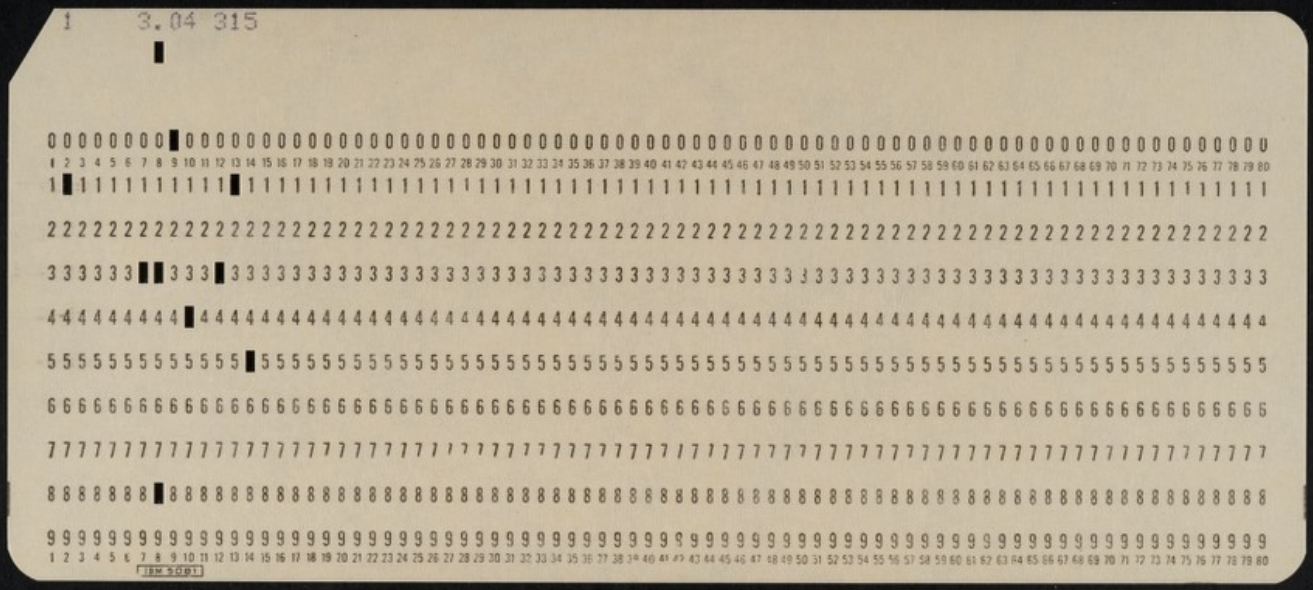

.0057
.0019
.0003
B/054 MOD
IBM 80/81

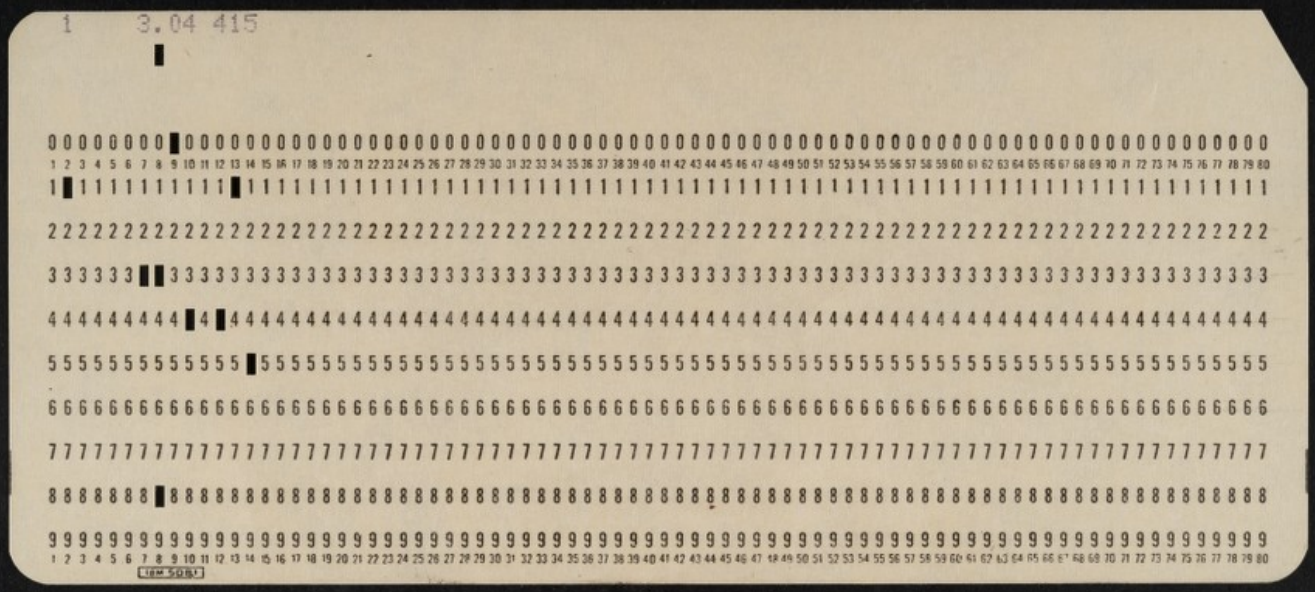

1 .1272 .0849 .0499 .0227 .0043 -.0047 -.0054 -.0010 .0041
IBM 5081

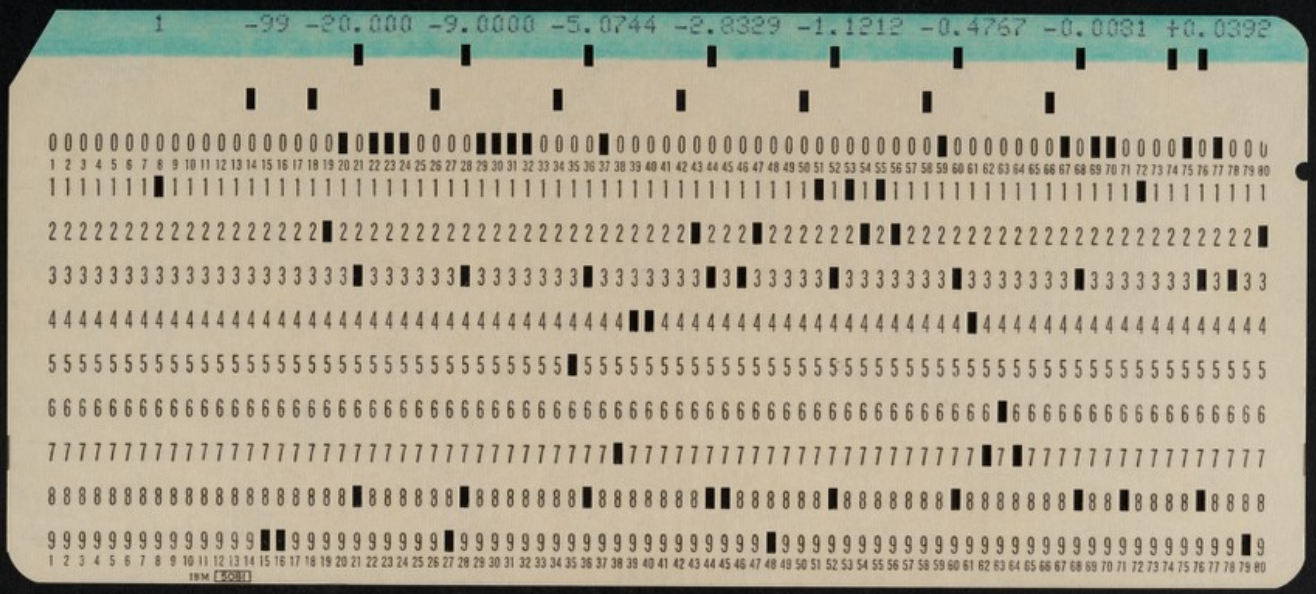

.0066
.0038
.0012
.0002
C/054 MOD
IBM 5081

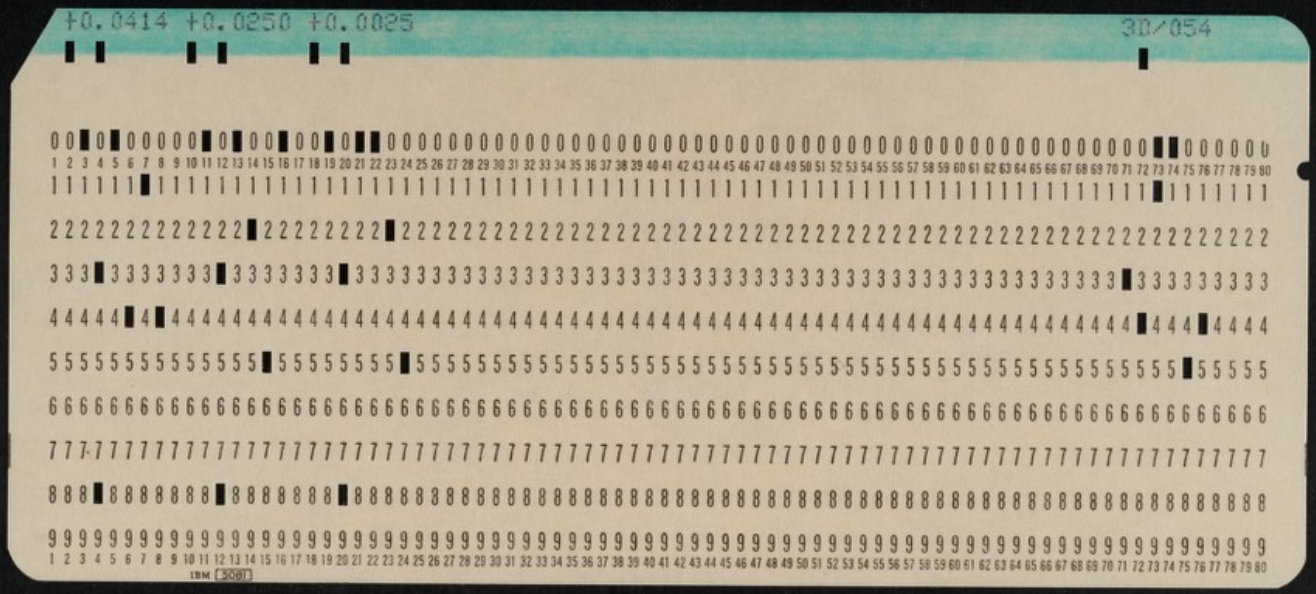

1 -99. -13.210 -7.857 -4.8774 -2.9142 -1.7673 -1.0394 -.5885 -.3244

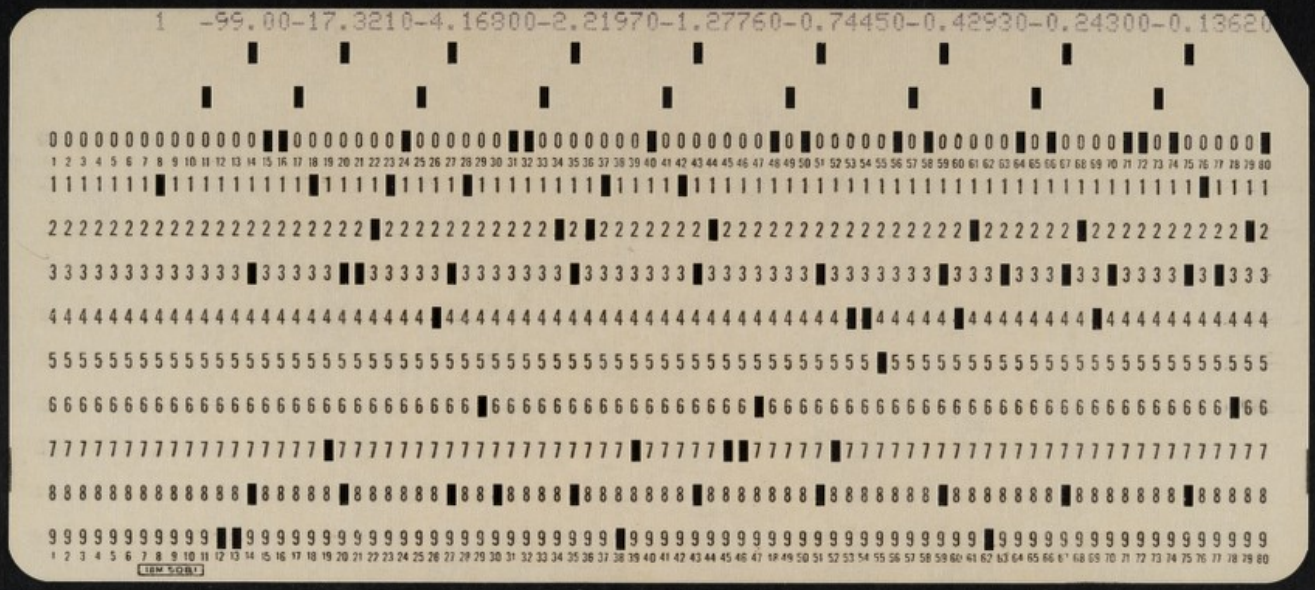

-.1512 -.0500 -.0060
D/054 MOD
IBM 5081

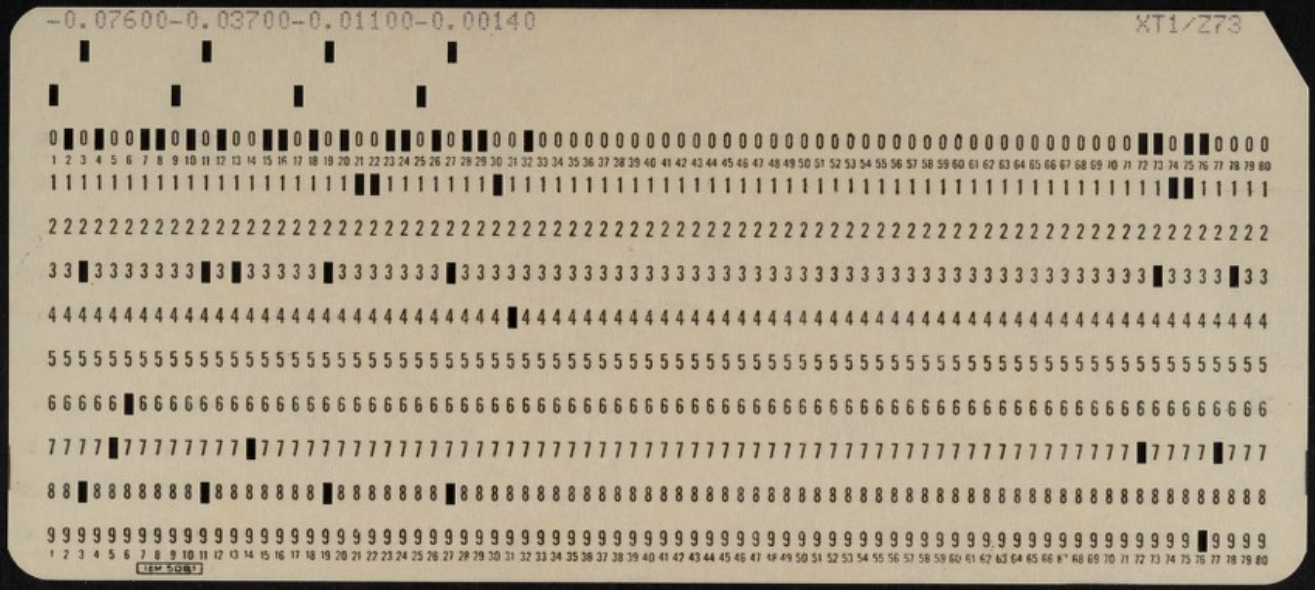

1 3.04 315
IBM 5081

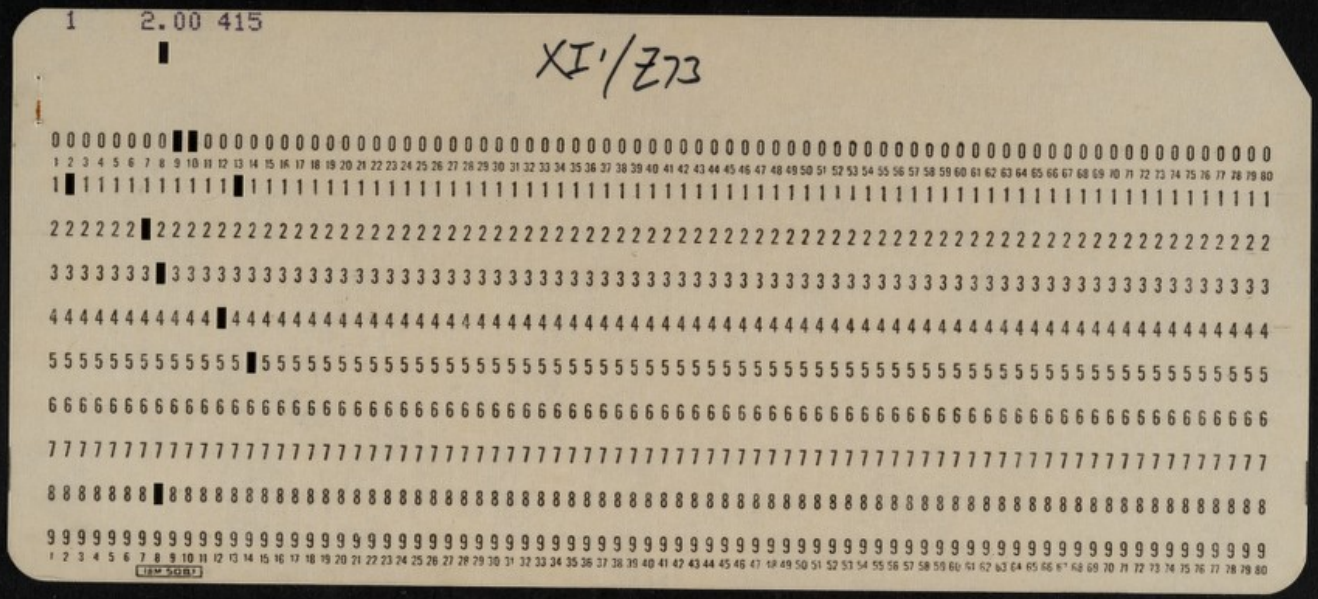

1 3.04 415

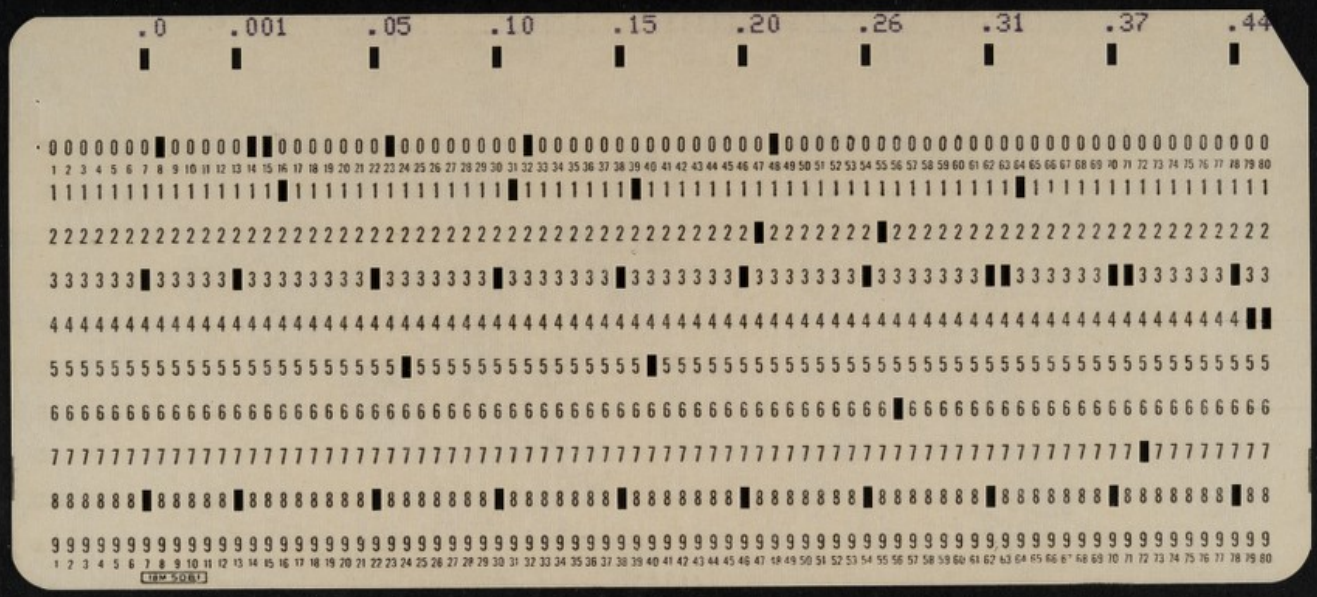

1 -99 -20.000 -9.0000 -5.0744 -2.8329 -1.1212 -0.4767 -0.0081 +0.0392
IBM 5081

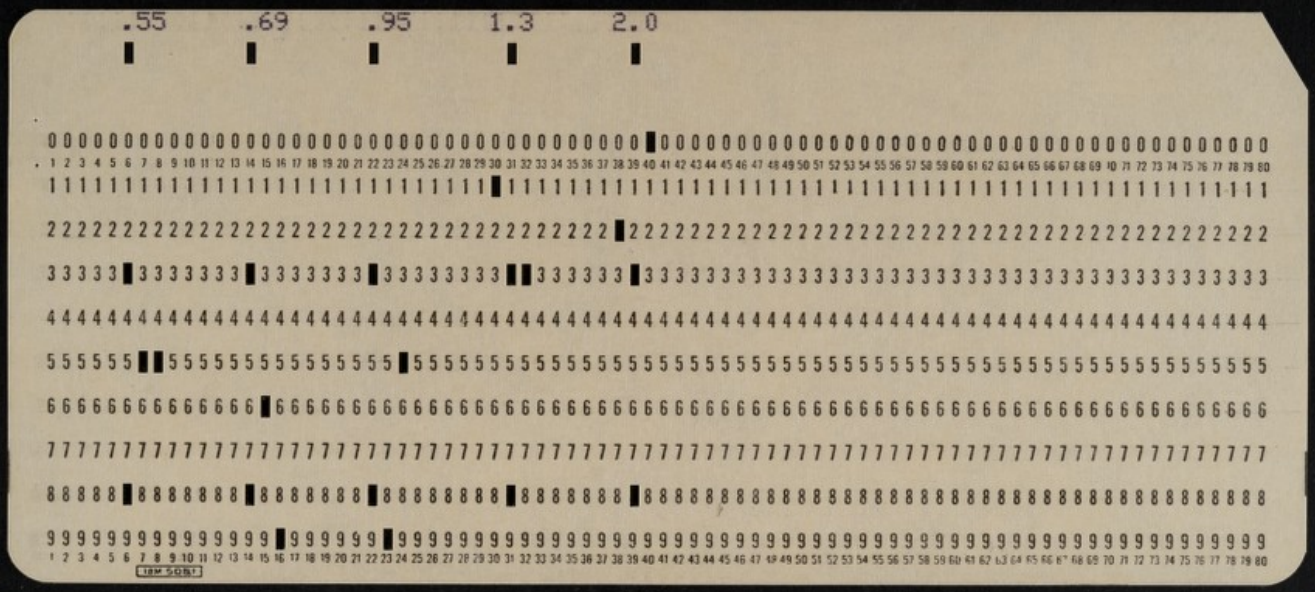

+0.0414 +0.0250 +0.0025
30/054
IBM 5081

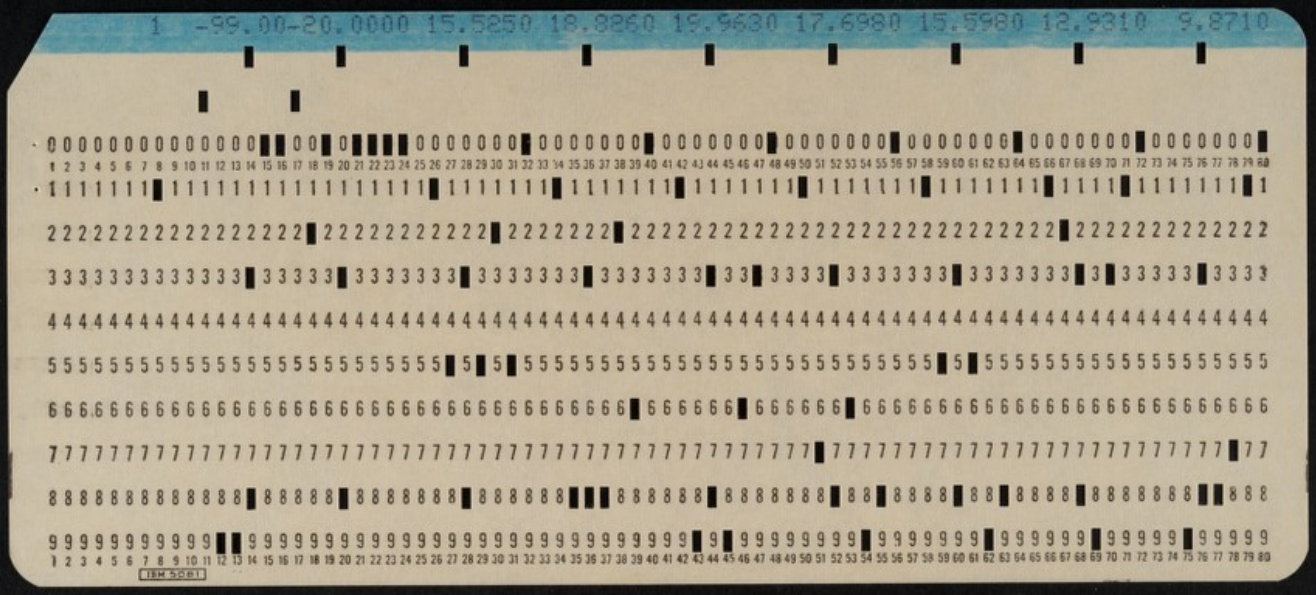

1 -99.00-17.3210-4.16800-2.21970-1.27760-0.74450-0.42930-0.24300-0.13620
IBM 5081

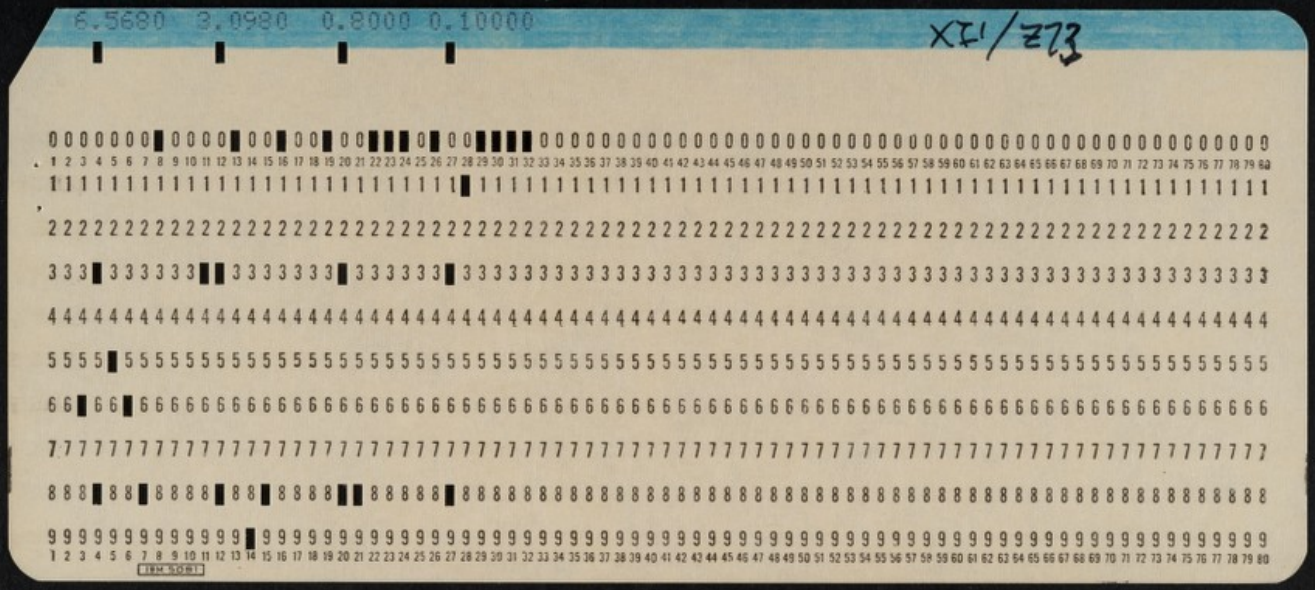

-0.07600-0.03700-0.01100-0.00140
XT1/Z73
IBM 5081

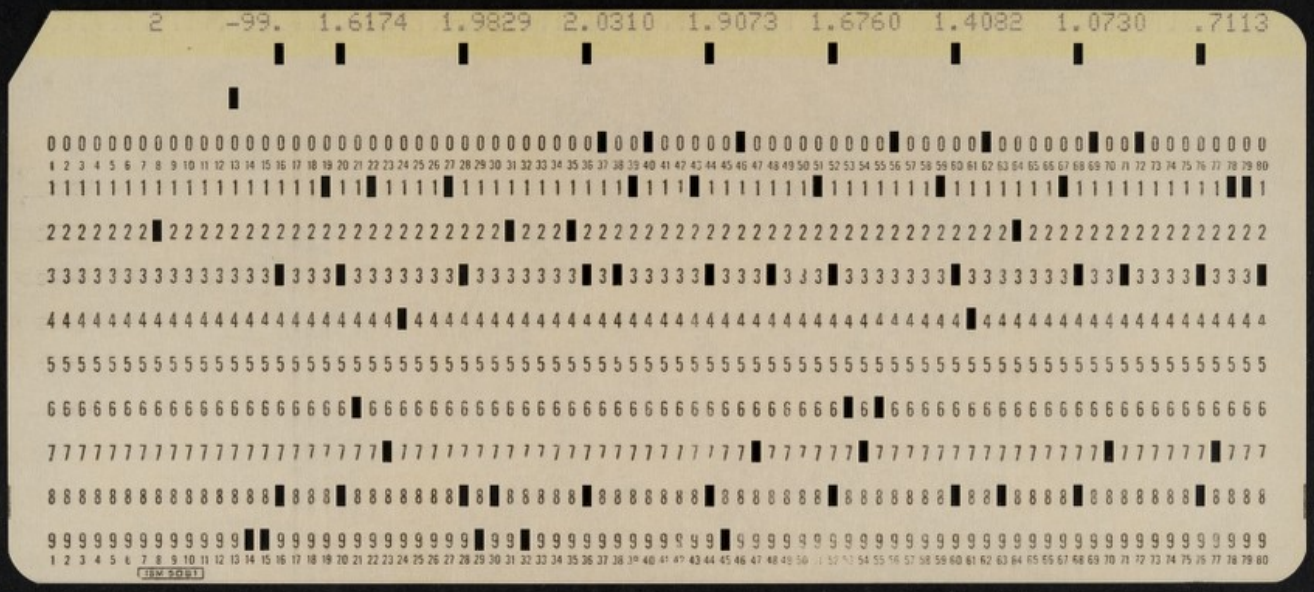

1 2.00 415
XI'/Z73
IBM 5081

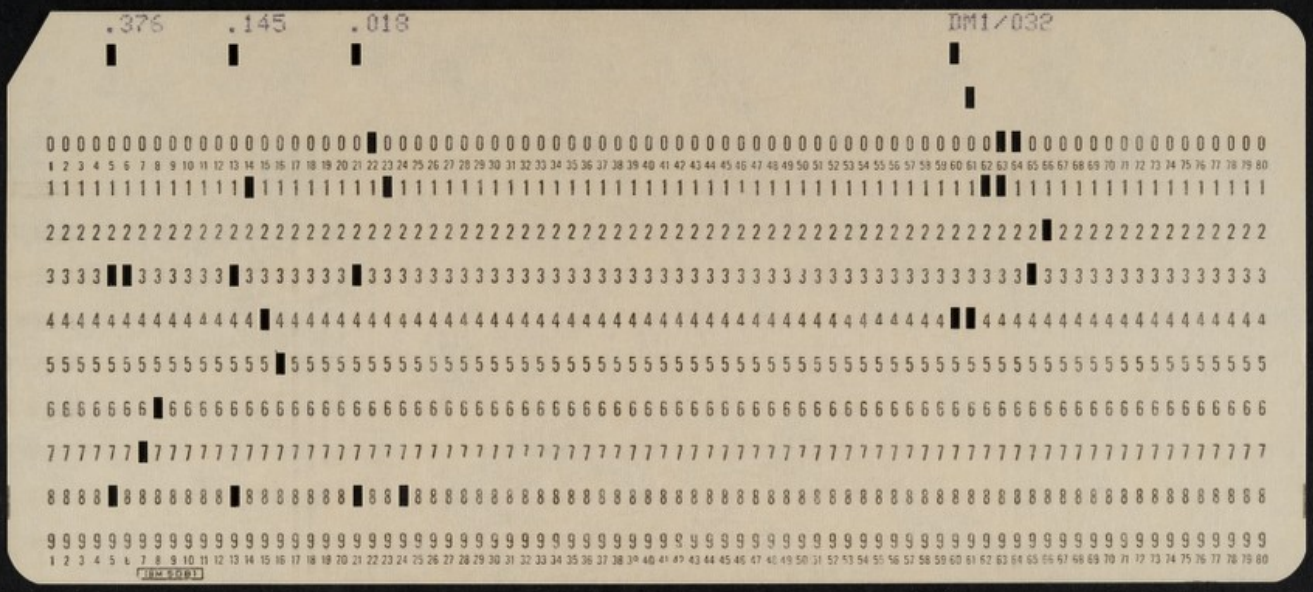

.0
.001
.05
.10
.15
.20
.26
.31
.37
.44
IBM 5081

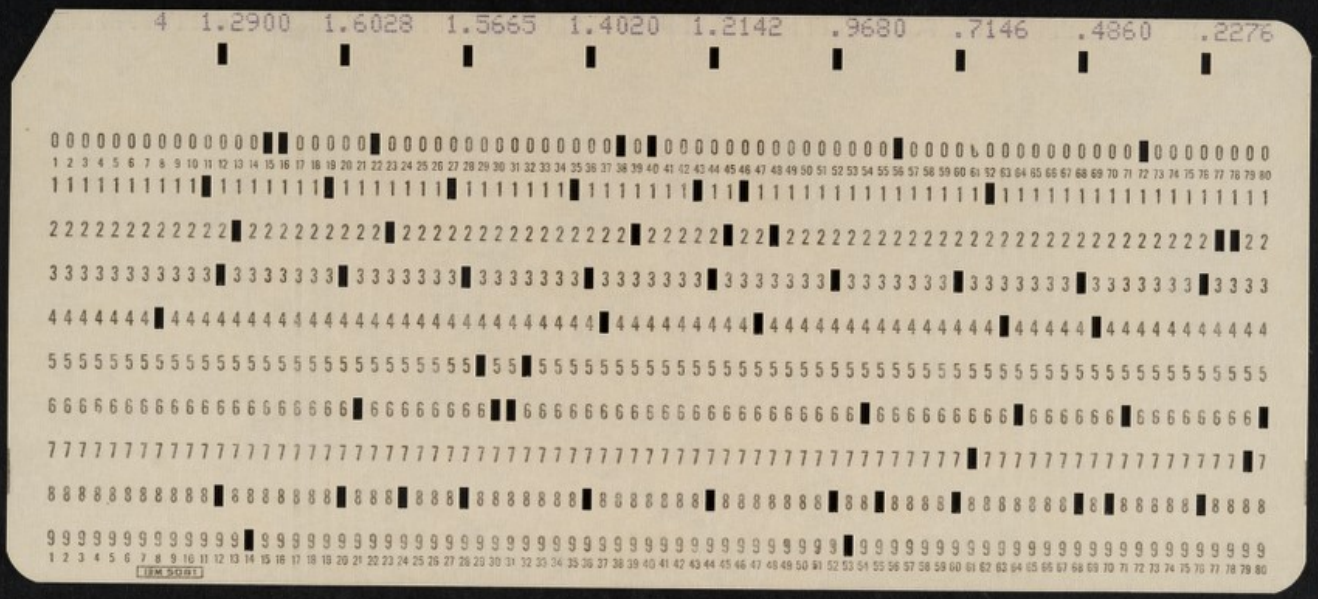

.55
.69
.95
1.3
2.0
IBM 5081

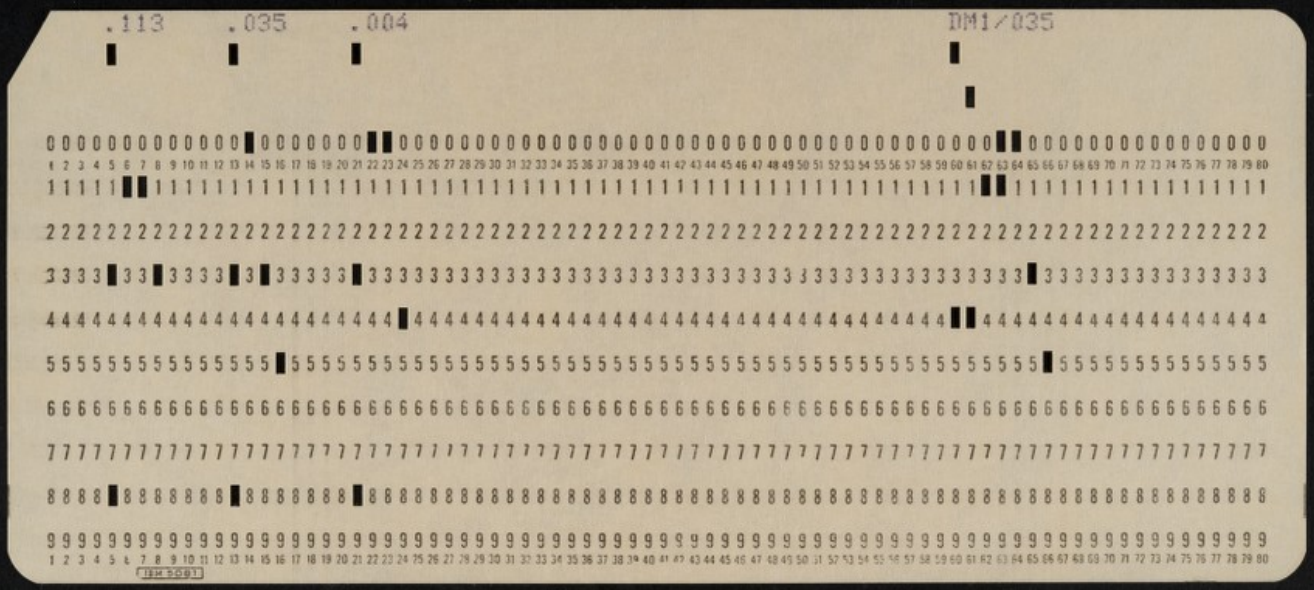

1 -99.00-20.0000 15.5250 18.8260 19.9630 17.6980 15.5980 12.9310 9.8710
IBM 5081

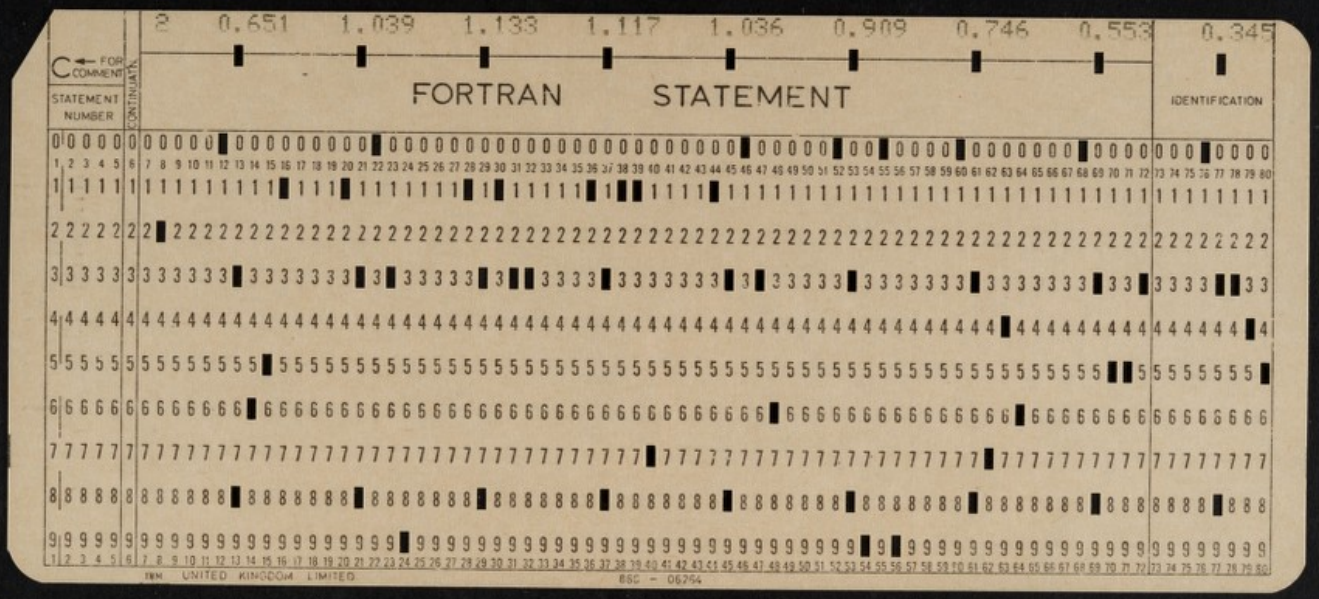

6.5680 3.0980 0.8000 0.10000
XI/273
IBM 5081

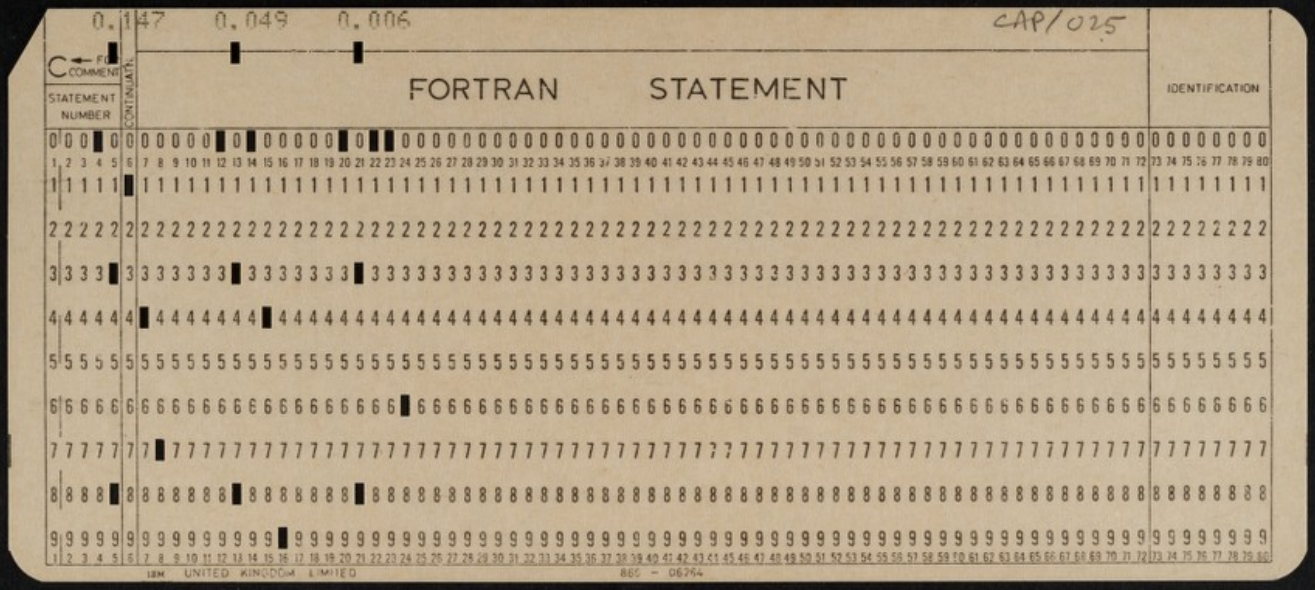

2 -99. 1.6174 1.9829 2.0310 1.9073 1.6760 1.4082 1.0730 .7113

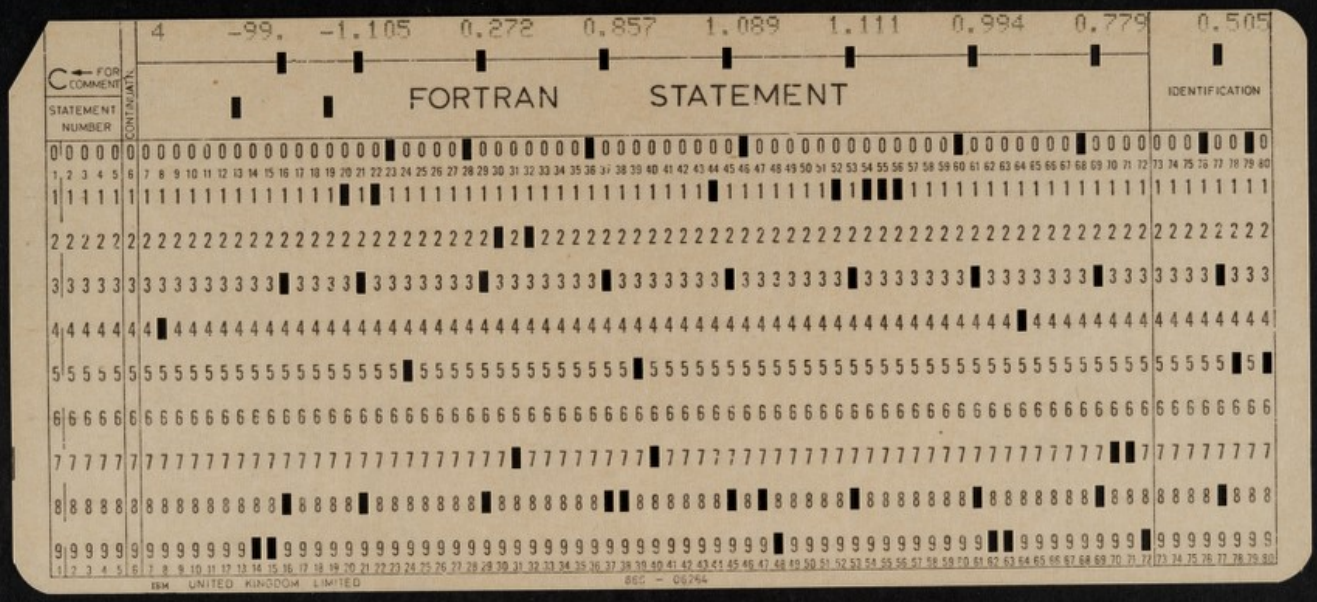

.376
.145
.018
DM1/032
IBM 5081

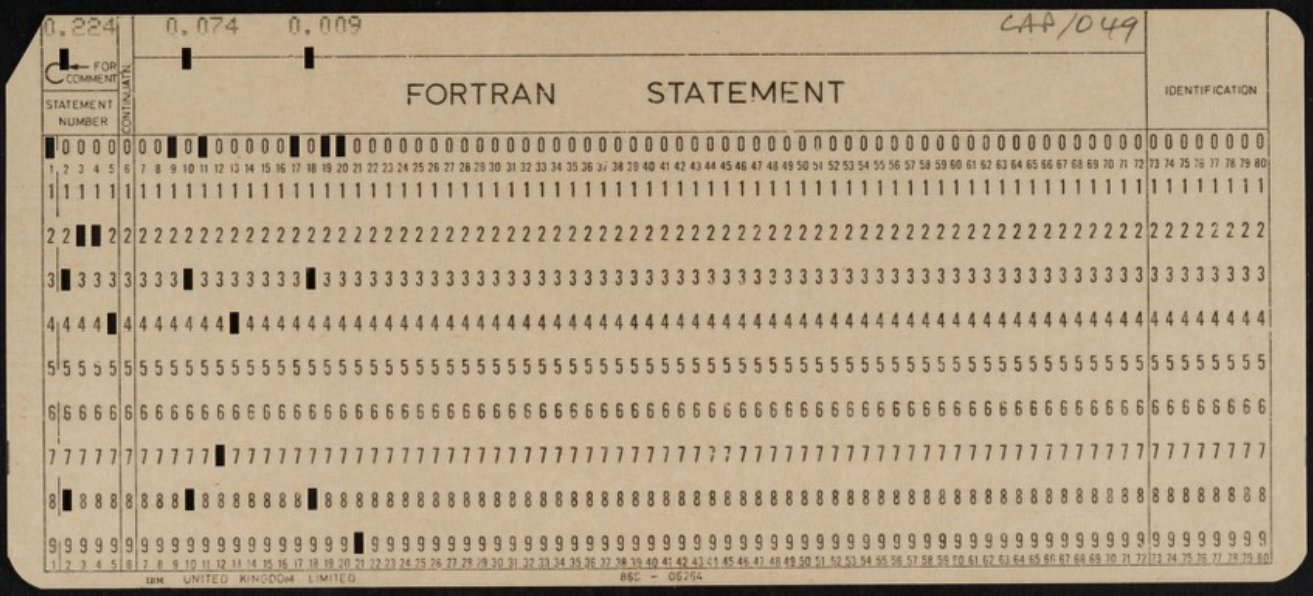

4 1.2900 1.6028 1.5665 1.4020 1.2142 .9680 .7146 .4860 .2276
IBM 5081

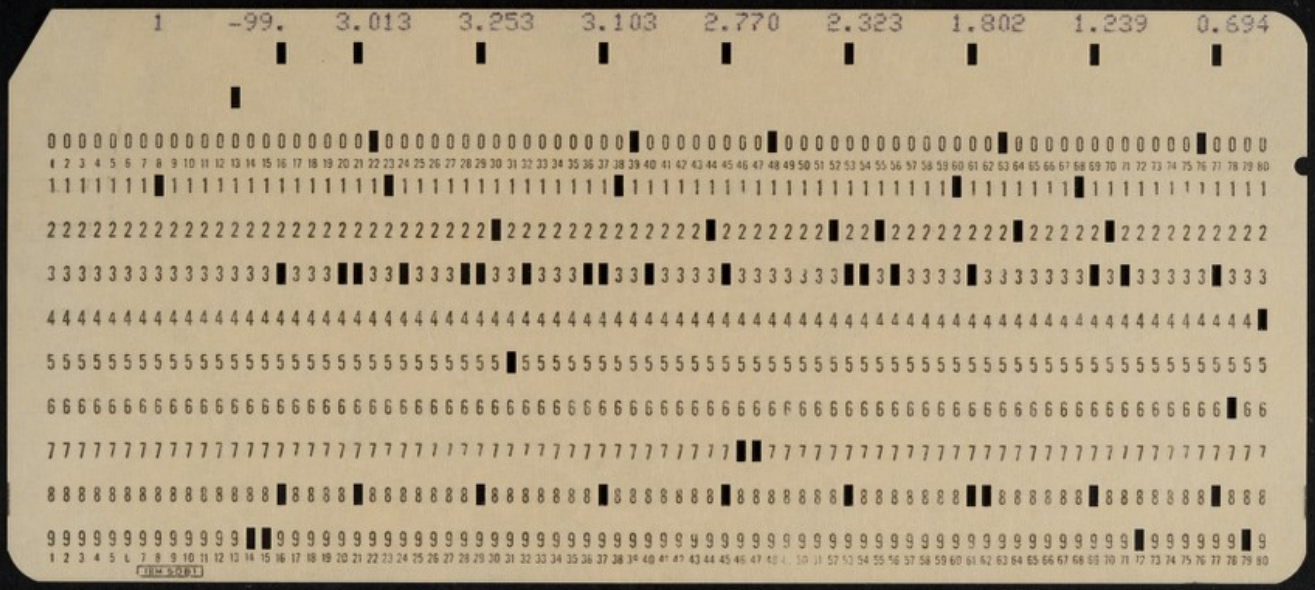

.113
.035
.004
DM1/035

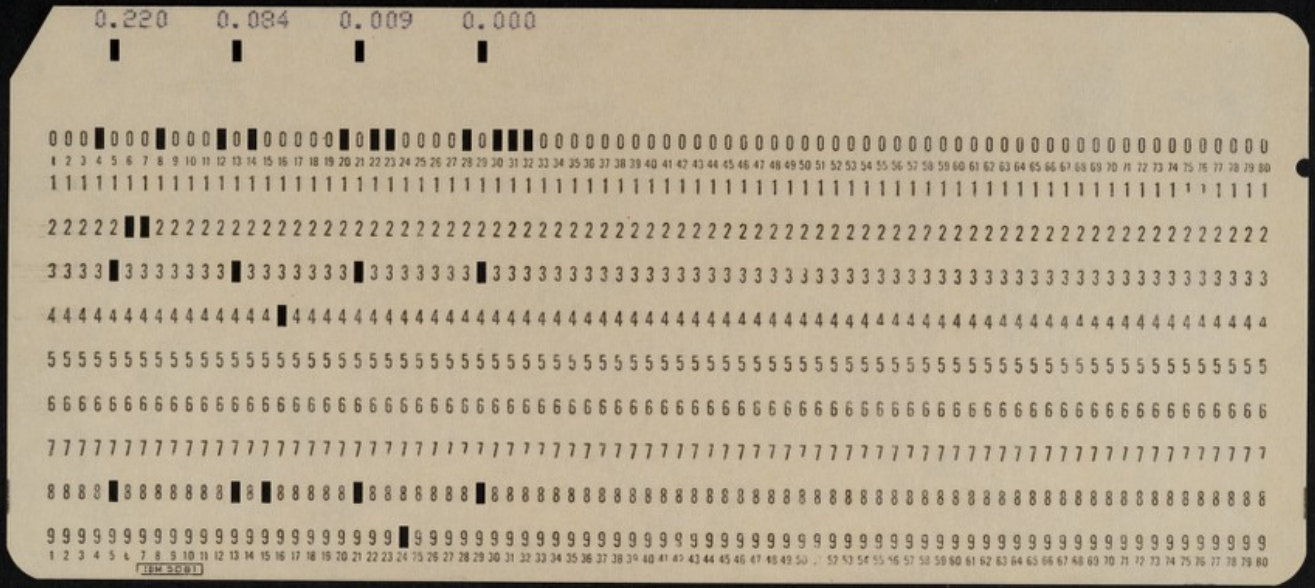

2 0.651 1.039 1.133 1.117 1.036 0.909 0.746 0.553 0.345
C ← FOR COMMENT
STATEMENT NUMBER
CONTINUATION
FORTRAN STATEMENT
IDENTIFICATION
IBM UNITED KINGDOM LIMITED
BSC - 06294


0.147
0.049
0.006
CAP/025
C ← FOR COMMENT
STATEMENT NUMBER
CONTINUATION
FORTRAN STATEMENT
IDENTIFICATION
IBM UNITED KINGDOM LIMITED
865 - 06264


4 -99. -1.105 0.272 0.857 1.089 1.111 0.994 0.779 0.505
C ← FOR COMMENT
STATEMENT NUMBER
CONTINUATION
FORTRAN STATEMENT
IDENTIFICATION
IBM UNITED KINGDOM LIMITED
860 - 08294

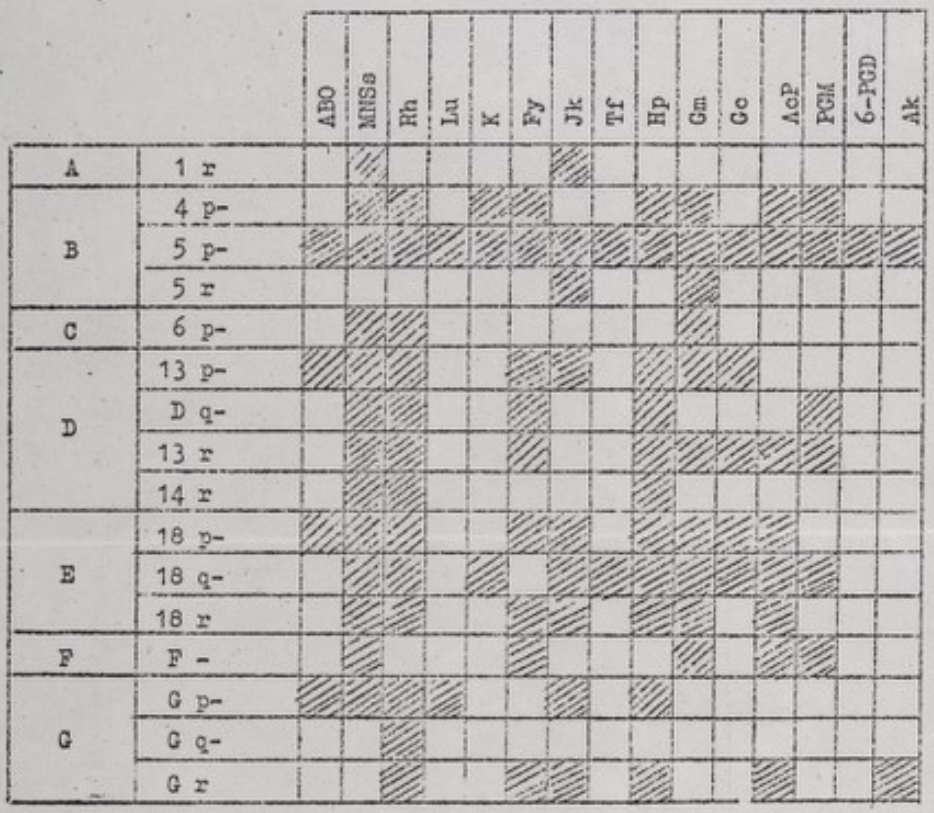

0.224
0.074
0.009
CAP/049
C FOR COMMENT
STATEMENT NUMBER
CONTINUATN
FORTRAN STATEMENT
IDENTIFICATION
IBM UNITED KINGDOM LIMITED
B60 - 05754


1 -99. 3.013 3.253 3.103 2.770 2.323 1.802 1.239 0.694


0.220
0.084
0.009
0.000

A provisional exclusion map - the by-product of attempts at deletion mapping.

LOCATIONS EXCLUDED



NOTE: A shaded area indicates that one patient (at least) is known to have the deletion and to be heterozygous at that locus. Due to individual variation in the extent of deletions, it is possible that a locus could be included in a large deletion and excluded in a small one.

12/IX/69

Departments of Child Health and Genetics, University of Glasgow.

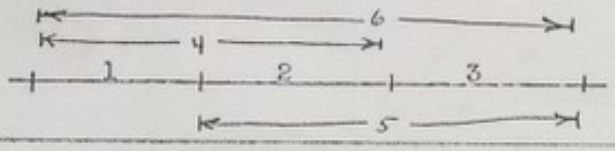
Ferguson-Smith. (Haigue.)

MAPIN

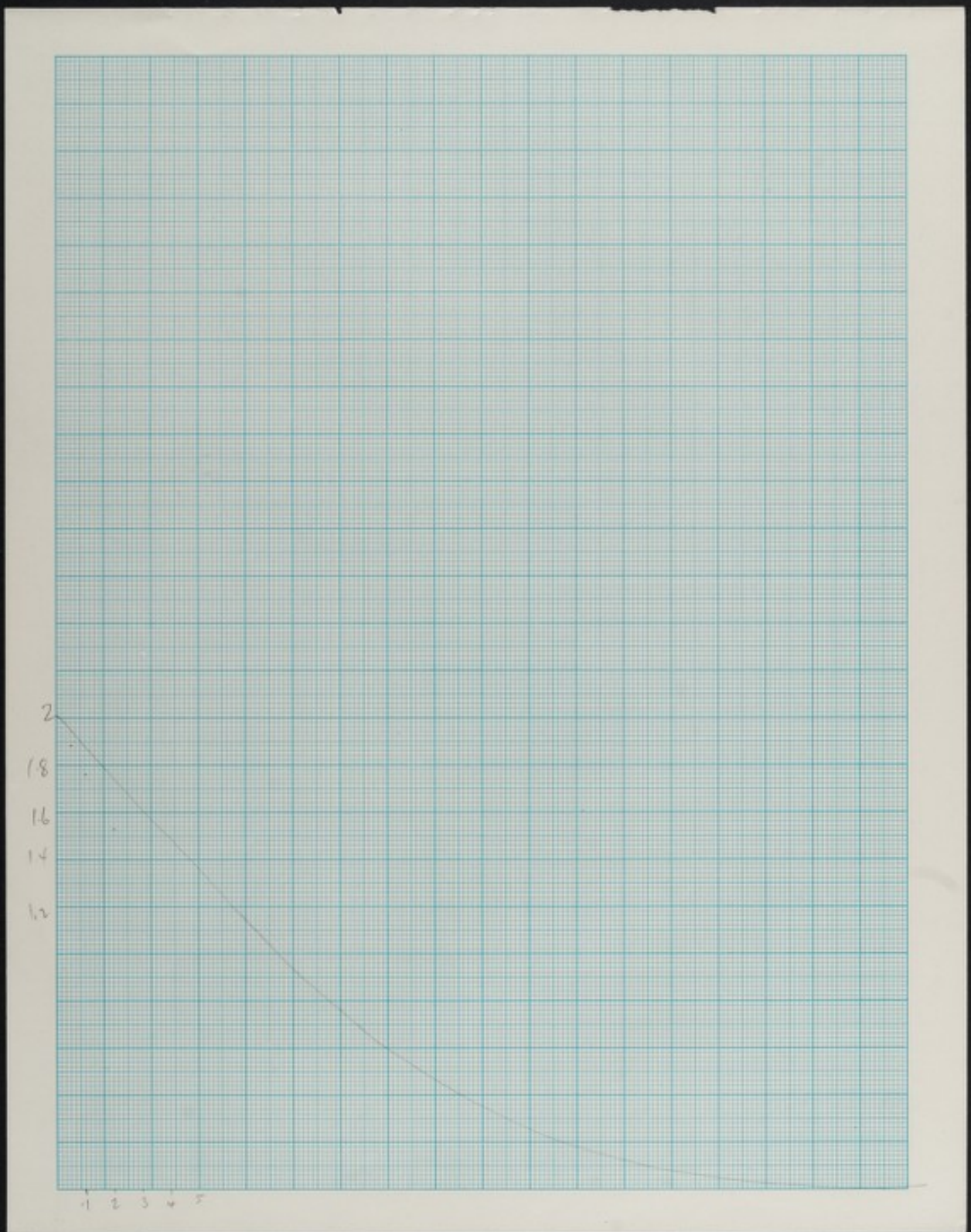
Map interval estimation program- Card input

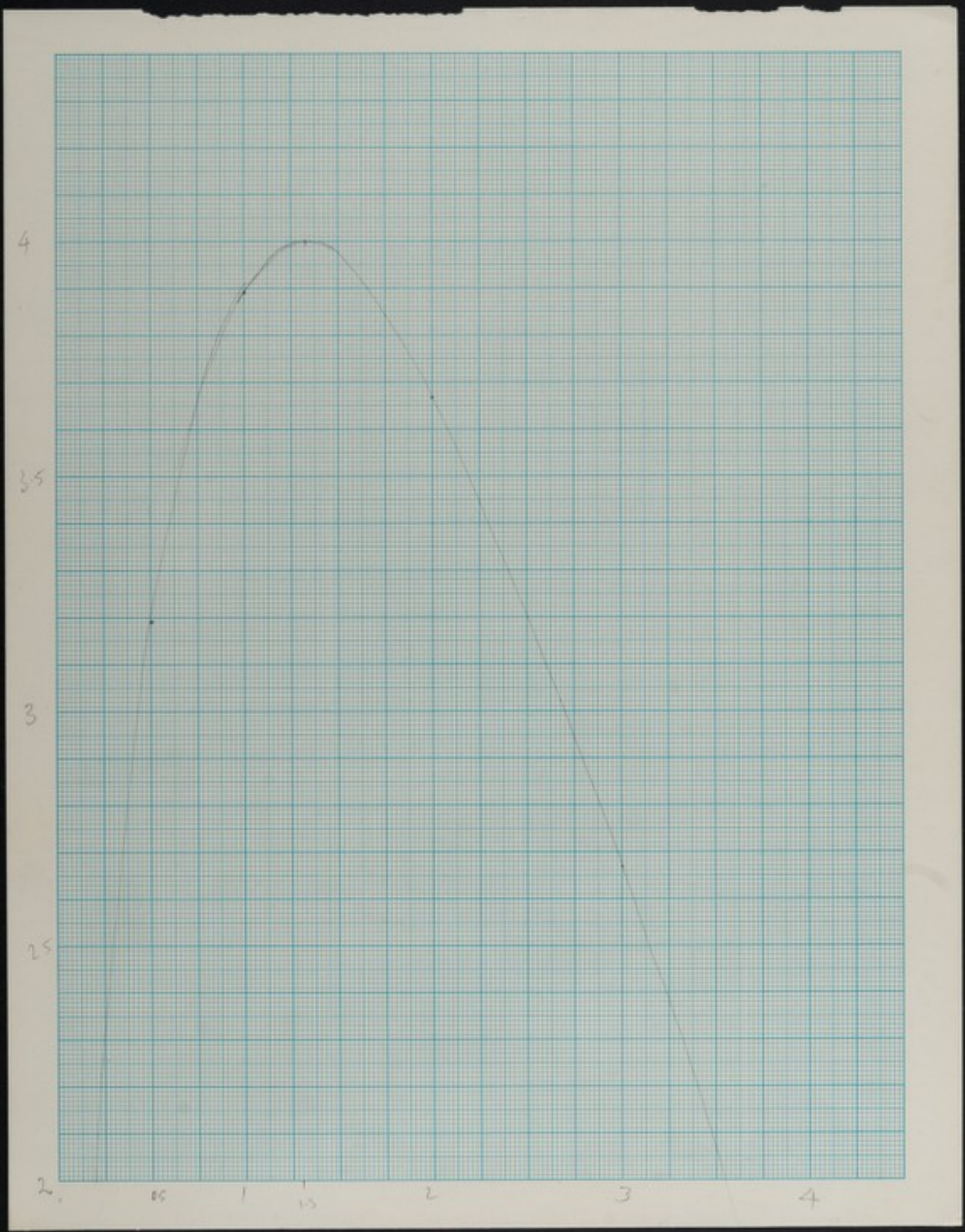
(All fields are right adjusted unless otherwise stated.)

Card or Group	Col	Description
1	1-2	KC- Number of chromosomes. This number is usually 22 or 1 depending whether the interval is being estimated on the autosomes or a particular chromosome such as the X.
	3-10	CL- The total length of the chromosome or chromosomes referred to by KC.
	11-12	NL- Number of loci. (3 or 4)
	13-14	NW- Number of points on each likelihood curve. All likelihood curves for each interval must have as input the lods for the same points.
2	10 fields of 8	W- Map values. (max 15) Additional cards are used when the number of map values exceeds 10.
3a	8	Map interval identification number. This will be a number 1 to 6 as defined by the interval diagram below. NEVER 3 FOR 3-LOCUS MAP.
	9 fields of 8 starting in col 9	Log likelihood values corresponding to the map values. When there are more than 9 values they are continued on cards with format 3b.
3b	10 fields of 8	This is a continuation of card 3a when there are more than 9 log likelihood values.
		(There may be 1 to 6 sets of number 3 cards. They need not be in any order and cards for a particular interval may be omitted when there is no information.)
4	8	This must be punched with a 9.



map interval diagram


2
1.8
1.6
1.4
1.2
1
2
3
4
5


4
3.5
3
2.5
2.
1
2
3
4

```
1      3.04 315                                    CHRØMØSØME 1
       0        .05        .10        .15        .20        .26        .31        .37        .44        .55
     .69        .95        1.5        2.0       3.04
       1        -99  -3.8259  -1.8159  -0.8319  -0.2945  -0.0136  +0.1063  +0.1277  +0.0966
+0.0416  +0.0136  +0.0017                                                                  1/054
1  2.84002 315                                    CHRØMØSØME 2
     .69        .95        1.4        1.8  2.84002
       1        -99  -0.0924  +0.8271  +1.1750  +1.2810  +1.2339  +1.0858  +0.8659  +0.6263
+0.3029  +0.0984  +0.0123                                                                  2/054

1  2.12045 315                                    2B CHRØMØSØMES
     .69        .95        1.2        1.5  2.12045
       1        -99  +0.1206  +0.2941  +0.3311  +0.3082  +0.2526  +0.1812  +0.1085  +0.0485
+0.0114  +0.0038  +0.0004                                                                  2B/054

1  1.65006 315                                    7C CHRØMØSØMES
     .69        .95        1.1        1.3  1.65006
       1        -99  -7.7136  -4.4857  -2.7793  -1.7085  -0.9975  -0.5222  -0.2168  -0.0426
+0.0320  +0.0200  +0.0020                                                                  7C/054

1  1.17013 315                                    3D CHRØMØSØMES
     .69        .95        1.0        1.05  1.17013
       1        -99  -20.000  -9.0000  -5.0744  -2.8329  -1.1212  -0.4767  -0.0081  +0.0392
+0.0414  +0.0250  +0.0025                                                                  3D/054
```

Green cards please

Urgent please

J Renwick

```
                PRIØRS FØR MAP INTERVAL, WRAW, ØN UNKNØWN AUTØSØME
Q       DN      32767

L       DRC     33.333,9.12,8.53,7.16,6.59,6.15,5.87,5.31,4.92,4.81,4.71,
                4.6,4.47,3.56,3.6,3.4,3.2,3.12,2.72,2.48,2.27,1.77,1.64
                    RELATIVE LENGTHS AS PERCENT TØTAL LENGTH (33.333MØRGANS)
W       DRN     16
LL      DRN     23
PW      DRN     16

WRAW    DRC     0,.05,.1,.15,.2,.25,.31,.37,.44,.55,.69,.95,1.5,2,3.04
                    RAW W IN MØRGANS. USE AS W/33.333 (PRØPØRTIØNAL W)

PAT     DBC     10,P(W) AT
MØR     DBC     10,MØRGANS

START   PPL

        LØØPX   1,=1,=1,=22
        RRM     L+X1,L+X1,LL+X1          SQUARED LENGTH =LL
        RRA     LL+X1                    SUM SQUARED LENGTHS (S)

        UNLØØP

        RRD     =2,,C                    -2/S
```

```
         LOOPX   1,=1,=1,=15
         LOOPX   2,=1,=1,=22
         RRD     WRAW+X1,L,W+X1      CONVERT RAW W TO PROPORTIONAL W

         JRR     L+X2,W+X1,UNL,UNL,  ADD L-W ONLY IF NON-NEGATIVE

         RRA     L+X2

         RRS     ,W+X1,PW+X1
```

```
UNL      UNLOOP

         RRM     C,PW+X1,PW+X1       COMPLETE P(W)

         RW      WRAW+X1,2,6
         BBR     6,,,Q,PAT,9
         BBR     7,MOR,,PAT,*+1
         BBR     1,==,1,PAT,*+1
         RWE     PW+X1,4
         BBR     Q,,,Q,PAT,*+1
         BP      PAT
         UNLOOP
         HLT
         END     START
```

chromosome 1. Prior odds = 1·0554

Let F = Duffy Fy
I = Uncoiler VII
M = Translocation point of t(1;16) FIM
T = " " " t(1;c) FIC

Order	Prob (relative) ×10³	But order is 'known' to be MIT
I F M T	·75	
F I M T	·63	
M F I T	·62	× ·62
M I F T	·45	× ·45
F I T M	·29	
I F T M	·27	
F M I T	·17	× ·17
I M F T	·15	
F M T I	·002	
M F T I	·002	
I M T F	·001	
M I T F	·001	

short arm: ·17, ·62 — long arm: ·45, 0

M I T

Fy on short : long

∴ ·79 : ·45 1·34

Prob 0·637 that Fy on short arm of I

if Fy on #1 (P_2 = 0·94)

Σ = 3·33 ×10³

$\bar{x}$ = ·2775 ×10³

$\Lambda' = \frac{277.5}{1.0554} = 262.9 : 1$

Prior 9·12 : 90·88

Final odds on assignment 2398 : 90·88 | 24·88·88

Prob 0·963

0 Σ = 1·24 ×10²

$\bar{x}$ = ·31 ×10³

$\Lambda' = \frac{310}{1.0554} = 293.7 : 1$

Prior 9·12 : 90·88

2678 : 90·88 | 2769·4

0·967.

Chromosome 16. length = 3.20% of $33\frac{1}{3}$M = 1.067M

let H = Hp locus (054)
1 = break-point of 1/16 translocation (F1M) (t(1;16))
2 = " " 2/16 " (F2M)
J = locus of polymorphic long 16 (JM1)

Order of loci (or reverse)				Relative, final odds $\times 10^4$		
1	H	2	J	·81		
2	H	1	J	·79		
H	2	1	J	·53	3.34	80
1	2	H	J	·52		
H	1	2	J	·36		
2	1	H	J	·33		
H	2	J	1	·17		
2	H	J	1	·16		
1	H	J	2	·04	0.43	9
H	1	J	2	·04		
1	2	J	H	·01		
2	1	J	H	·01		

$\Sigma = 3.76 \times 10^4$

$\bar{x} = 0.31363 \times 10^4$

$\Lambda' = \frac{\bar{x}}{\text{prior}} = \frac{0.31363 \times 10^4}{1.03501} =$ Syntany 16 : asyntany 3030.2 : 1

Prior 3.2 : 96.8
Final 9696.64 : 96.8
or 100 : 1 — as expected from band calculation

Assurance = 0.99.

If so, 90% probability that J does not lie among the other 3 points.

A = Fy Fy
B = V11 U
C = V12 I
D = FIC T

$\frac{23}{45}$

SUMMARY OF RESULTS

ORDER	143	BACD	UFIT .231945E004	0.097 (0.011 TO 0.231)	0.000 (0.000 TO 2.320)	0.463 (0.000 TO 2.372)
ORDER	123	ABCD	FUIT .223334E004	0.097 (0.011 TO 0.230)	0.000 (0.000 TO 2.335)	0.373 (0.000 TO 2.363)
ORDER	415	CABD	IFUT .178218E004	0.000 (0.000 TO 2.274)	0.099 (0.011 TO 0.234)	0.665 (0.244 TO 2.495)
ORDER	216	CBAD	IUFT .153112E004	0.000 (0.000 TO 2.301)	0.096 (0.011 TO 0.228)	0.757 (0.304 TO 2.509)
ORDER	153	ABDC	FUTI .151215E004	0.099 (0.011 TO 0.234)	0.656 (0.242 TO 2.493)	0.000 (0.000 TO 2.332)
ORDER	163	BADC	UFTI .140183E004	0.097 (0.011 TO 0.231)	0.753 (0.301 TO 2.508)	0.000 (0.000 TO 2.330)
ORDER	425	ACBD	FIUT .277079E003	0.000 (0.000 TO 0.179)	0.000 (0.000 TO 0.197)	0.682 (0.238 TO 2.501)
ORDER	246	BCAD	UIFT .254568E003	0.000 (0.000 TO 0.194)	0.000 (0.000 TO 0.176)	0.786 (0.311 TO 2.518)
ORDER	435	ACDB	FITU .125966E001	0.000 (0.000 TO 2.246)	0.347 (0.000 TO 2.322)	0.445 (0.069 TO 2.393)
ORDER	465	CADB	IFTU .945429E000	0.000 (0.000 TO 2.177)	0.624 (0.110 TO 2.428)	0.406 (0.019 TO 2.321)
ORDER	236	BCDA	UITF .871262E000	0.000 (0.000 TO 2.275)	0.000 (0.000 TO 2.273)	0.675 (0.256 TO 2.502)
ORDER	256	CBDA	IUTF .770439E000	0.000 (0.000 TO 2.258)	0.402 (0.017 TO 2.308)	0.616 (0.105 TO 2.422)

WEIGHTS OF LOCI POSITIONS

0.509 1.411 1.524 0.623

.902 — .902

.509 .622 — .623

V11 V12

$\frac{1}{5}$ $\frac{1}{5}$

V11 .097 Fy 1 A V12 .463 3 FIC

V11 V12 Fy FIC
B 2 C 4 A 6 D

1×10^4

23 : 22 : 18 : 15 : 15 : 14 : 3/3

~~78~~ 84

If V11 = U is at 2 constriction and q = prob of Fy lying between U (V11) and T (FIC)

p = prob of Fy not

$q : p = [.23 + .14 + .03] : [.22 + .18 + .15 + .03]$

$= .55 \quad : .58$

Prior distributions of map intervals, between 2 of n loci.

n=1 Assume n+1 events on a circle, 1 being the opening-up event.

$p(w) \propto 1/L$ (i.e. distance from locus to end is uniformly distributed at density 1/L.)

n=2 Imagine an extra event on this circle.

$p(w) \propto (1/L)\frac{(L-w)}{L}$ where w is the distance from end and does not contain the third locus. However, owing to symmetry of 3 events on a circle, this end interval is distibuted exactly as the interval between any two neighbouring events out of 3.

$p(w \text{ inner}) \propto (L-w)/L^2$ i.e. interval between loci or interval to near end.

$p(w \text{ outer}) \propto w/L^2$ i.e. from locus to distant end.

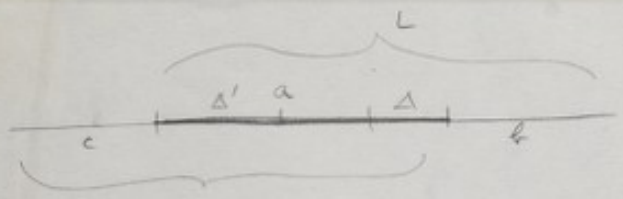
n=3 As for n=2 but combined with probability of extra event falling outside the interval.

Prior prob of a particular sequence is 1/3. Given a specific sequence, $w_1+w_2=w_3$

$p(w_1 \text{ inner}) \propto (L-w_1)^2/L^3$

$p(w_3 \text{ outer}) \propto w(L-w_3)/L^3$

$a > b > c$



$$\frac{L}{2} + \frac{\Delta}{2} = \frac{L'}{2} + \frac{\Delta'}{2}$$

Trains in a, $p(\omega) = \dfrac{2(L+\Delta)-4\omega}{T(L+\Delta)}$ or $\dfrac{2(L'+\Delta')-4\omega}{T(L'+\Delta')}$

Trains in b,

$$p(\omega) = \frac{2\omega}{T(L+\Delta)} \qquad \omega < \frac{L}{2} - \frac{\Delta}{2}$$

$$p(\omega) = \frac{(L-\Delta)}{T(L+\Delta)} \qquad \frac{L}{2} - \frac{\Delta}{2} < \omega < \frac{L}{2} + \frac{\Delta}{2}$$

$$p(\omega) = \frac{2(L-\omega)}{T(L+\Delta)} \qquad \frac{L}{2} + \frac{\Delta}{2} < \omega < L$$

0 elsewhere

Trains in c

$$p(\omega) = \frac{2\omega}{T(L+\Delta)} \qquad \omega < \frac{L'}{2} - \frac{\Delta'}{2}$$

$$p(\omega) = \frac{L'-\Delta'}{T(L+\Delta)} \qquad \frac{L'}{2} - \frac{\Delta'}{2} < \omega < \frac{L'}{2} + \frac{\Delta'}{2}$$

$$p(\omega) = \frac{2(L'-\omega)}{T(L+\Delta)} \qquad \frac{L'}{2} + \frac{\Delta'}{2} < \omega < L'$$

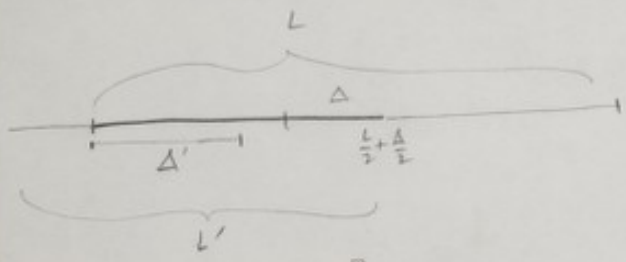
0 elsewhere

$\sum$ at $0 = \dfrac{2}{T}$

at $c = \dfrac{2(L'+\Delta') - 2(L'-\Delta') + 2(L'-\Delta')}{T(L+\Delta)}$

$= \dfrac{2}{T}$

at $b =$

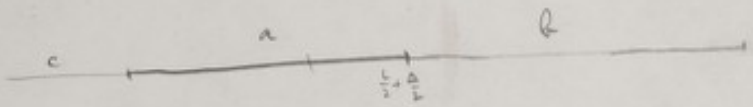


Soit $0 \quad p(w) = \frac{2}{L}$

$\frac{L-\Delta}{2}$

$T =$

$a > b > c$

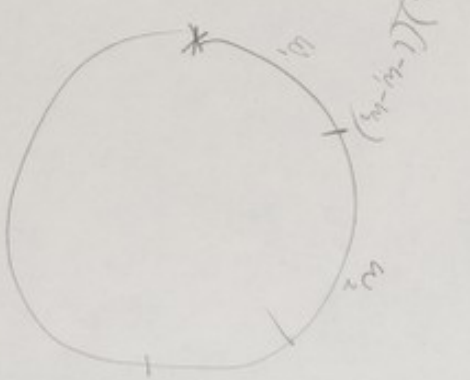


$$L + \Delta = L' + \Delta'$$

$$T = L + L' - \frac{L}{4} - \frac{\Delta}{4} - \frac{L'}{4} - \frac{\Delta'}{4} = \frac{1}{4}\left[3L + 3L' - \Delta - \Delta'\right]$$

$$= \frac{1}{4}\left[2L + 2L' + 2(L\right.$$

$$T = \frac{1}{2}\left(L + L' + (b + c)\right)$$



$a_l + a_s = t_l + t_s = \Sigma$

~~$a_l - t_l = t_s - a_s$~~

$t_l - a_l = a_s - t_s$

$t_l = a_s + a_l - t_s$ $\qquad t_l = \Sigma - t_s$

$a_s + a_l - a_s - a_l + t_s$

---

(1) o-s.

(2) From $\frac{A}{2} - 2s - \delta$ to $\frac{A}{2} - s - \delta$

A

let A = sum of all 4 arm-lengths
s : shortest of all 4 arm-lengths.
δ = change in length due to translocation

Limits of break-point are

1. On shortest arm
2. On other chromosome arm $\left(\frac{\Sigma}{2} - 2s - \delta\right)$ to $\left(\frac{\Sigma}{2} - s - \delta\right)$

t(16q+ ; 1p-)