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PATTERNS OF INHERITANCE

PHENOTYPE EXPRESSION
IN
ONE FAMILY TREE

[an early CPA BookClub Booklet 1991]

ABSTRACT

Gregor Mendel described the results of over two decades of research concerning the traits, forms, hybridization, and statistics of his breeding experiments with the garden pea, Pisum spp. to the Natural History Society of Brünn in 1865. These results appeared in print in 1866 and 1869. He had followed and recorded generations of offspring and had come to the conclusion that there were pure traits and hybrid traits and that in the hybrid traits, there were dominant and recessive characteristics which fell into a mathematical relationship of 3:1 (dominant to recessive). Through further experimentation, Mendel found that each successive generation adhered to the same ratio and that the recessive trait would sometimes appear after skipping a generation. As the study of genetics progressed, it was found that these experiments and ratios could be associated with any reproductive animal or human being and remained the same. Positive and negative (diseases) traits or phenotypes could be followed from past observations and tested and treated so that they could or could not occur again. The purpose of this paper is to observe the phenotypes and test the 3:1 ratio in several generations of one family to see if the ratio holds true.

The phenotypes found within a family tree, going back 14 generations to 1623, were looked at and the dominant and recessive traits within 31 families dating from 1750 were found to adhere to the 3:1 ratio in the pedigree analysis.

CURRENT GENETIC DEVELOPMENTS

The present knowledge about genetics through the works of scientists in this Century begins with the work of Carl Correns (1900) et al. and Walter Sutten (1902) (Penny, 1965; Raven, 1988) who, after rediscovering the work of Mendel on inherited traits (Raven, 1988), developed theories concerning inheritance through chromosomes. In 1910 the geneticist, T.H. Morgan, determined that chromosomes were sex linked through his experiments involving fruit fly (Drosophila) (Raven, 1988) and after his Mechanism of Mendelian Heredity (1915) and The Physical Basis of Heredity (1919) (Penny, 1965) were published, his chromosome theory of inheritance became the basis for modern genetic research. The next major scientific step occurred in 1931 when C. Stern observed that genes assorted themselves differently on the chromosomes and that there was a crossing over during meiosis (Raven, 1988).

All of this research was based on the work of Gregor Mendel (1822-1884), who followed the generations of Pisum spp. and recorded the variances, numbers of each variance and ascertained the statistical relationships of the results (Penny, 1965). Mendel learned four things in regards to heredity: 1) the traits that he studied did not produce intermediate types when crossed but instead inherited the distinct characteristics that were either seen or not seen in the true generation, 2) for every pair of traits observed, one did not appear in the F1 generation but appeared in the F2 generation, 3) pairs of alternative traits segregated themselves amongst the offspring (some exhibiting one trait, some the other),

4) The traits that occurred in the F2 generation, were expressed in a ratio of 3/4 dominant and 1/4 recessive (3:1) (Raven, 1988).

Mendel developed a model, based on what he had learned in regards to heredity. The model (Raven, 1988) consists of five components:

A) Parents do not transmit physiological traits directly to their offspring, rather, transmitting information about the traits. [Mendel called these traits - factors. ed.]

B) In diploid (having two sets of chromosomes) organisms, each parent has two `factors'. If they are different, they are heterozygous and if they are the same, they are called homozygous.

C) The alternative forms of a `factor' or gene leading to alternative character traits is called an allele.

D) Two alleles, one from the male gamete and one from the female gamete [fusion of cells through fertilization ed.] remain distinct and do not blend with each other. The gametes of the mature offspring have equal proportions of the genetic information received from each parent.

E) A particular encoded gene trait does not ensure that trait will be expressed. Only one allele (the dominant one) is expressed while the other allele (recessive) is present but unexpressed. The total set of genes present in the cells is called the genotype while the actual physical expression of the cells is called the phenotype.

Various traits in human beings display the same dominant and recessive inheritances found in the experiments performed by Gregor Mendel. Haemophilia, albinism, tallness and shortness, color of skin and hair, Tay-Sachs disease, Cystic-fibrosis, Arthritis and length of life are just a few of the dominant and recessive traits found in human beings. Many of the phenotypes that one sees were there before birth in the chromosomes of the parents: the color of the skin and eyes, similar features as one of the parents, and future similarities (baldness, Wardenberg syndrome etc) of the parents that are recessive.

The subjects dealt with in this paper are represented by 31 families (ALLEN: BARDINI: CARR: COTNEY: DAILEY: ELBERT: ENGLAND: FUNDERBURK: GAITHER: GAY: HEFLIN: HENSON: KILGORE: MANN: MAY: MCELWEE: MITCHELL: MOTLEY: MUNS: PATRICK: PENZ: PHILLIPS: RAY: ROCKETT: RUNYAN: SAMFORD: SHERIDAN: TRAVIS: WALKER: WALMSLEY: WOOD) spanning the time period from 1750 - 1991, obtained from several family trees that extend back to 1630 in the southern USA and Canada. Where there were no photographs or drawings available, prior to 1875, the information was obtained from historical records and personal interviews (Scarborough, 1991; Runyan-Wright, 1980). At last count (Scarborough, 1991) there were over 300 living members of the Gay family and 400 members of the Runyan family alone. For this paper, a random sampling of 135 members representing all the above families from the 9th Generation to the 13th Generation were used.

The following dominant and recessive phenotypes were looked at; Tallness, shortness, baldness, right semi-cauliflower ear, dark hair, light hair, premature greying, middigital hair, longevity, haemophilia, and arthritis.

T = Tall 5'8" + ♀ & 6'+ ♂, t = <5'6"
L = Light hair (dominate) b (recessive)
D = Dark hair d ibid:
B = Bald b ibid:
M = Middigital hair m ibid:
O = longevity age 75-115 at time of death
o ibid:
H = Haemophilia
P = Premature greying p ibid:
A = Arthritis a ibid:
R = Right semi cauliflower ear r ibid:

Pure Generation XI

D↓ R→	t	b	l	d	m	p	a	o
T	Tt	Tb	Tl	Td	Tm	Tp	Ta	To
B	Bt	Bb	Bl	Bd	Bm	Bp	Ba	Bo
L	Lt	Lb	Ll	Ld	Lm	Lp	La	Lo
D	Dt	Db	Dl	Dd	Dm	Dp	Da	Do
M	Mt	Mb	Ml	Md	Mm	Mp	Ma	Mo
P	Pt	Pb	Pl	Pd	Pm	Pp	Pa	Po
A	At	Ab	Al	Ad	Am	Ap	Aa	Ao
O	Ot	Ob	Ol	Od	Om	Op	Oa	Oo

Ninth Generation Chart of F1 offspring born between 1860 - 1899
Example: (T) Tall where T is (tall pure) dominant and t is (short pure) recessive.

F1	t	t
T	Tt	Tt
T	Tt	Tt

All offspring will exhibit signs of Tall. Any other crossing will give the same results for F1 generation, Middigital hair, Arthritis, Premature greying etc. When the F1 Generation is allowed to breed, the ratio of dominate to recessive genes occurring in the offspring will be 3:1.
Example: from the above chart:

F2	T	t
T	TT	Tt
t	Tt	tt

If four offspring are produced in the F2 Tenth Generation then three will exhibit the trait of Tall and the fourth will not. One quarter of the F2 Generation will be pure bred Tall, one half will be hybrid tall and one quarter will be pure bred not tall. Therefore, on the average, there will be three persons with tallness and one not, in the F2 Generation for a ratio of 3:1.
If the F2 Generation is allowed to breed and form the F3 Eleventh Generation, The pure tall (TT) will only produce tall offspring, the pure short (tt) will produce only short offspring and the hybrid tall (Tt) will produce offspring in the ratio of 3:1.

Example of the F3 Generation:

F3	TT	Tt	Tt	tt
TT	TTTT	TTTt	TTtT	TTtt
Tt	TTTt	TTtt	TtTt	Tttt
Tt	TtTT	TtTt	ttTT	ttTt
tt	TtTt	Tttt	ttTt	tttt

F3 Generation Phenotype Ratio

PHENOTYPE	GENOTYPE	P RATIO
Pure tall	TTTT(1) TTTt(2) TtTT(2) TtTt(4)	9
Hybrid tall	TTtt(1) Tttt(2)	3
Hybrid tall	ttTT(1) ttTt(2)	3
Non tall	tttt	1

Genealogical Accountability Table (GAT)
T = Tall
S = Short
L = Light hair
D = Dark hair
B = Bald
M = Middigital hair
O = Longevity 75-115 at time of death
o = 50-75 at time of death
H = Haemophilia
P = Premature greying
A = Arthritis
R = Right semi cauliflower ear

GAT # OF PERSONS RATIO # RATIO OF D:R

T	95	23.75	3:1
S	37	9.25	↑
L	38	9.5	↓
D	115	29	3:1
B	8	2	↑ 16:1
M	11/3	2.75:.75	3:1
o	14	3.1	↓
O	44	11	3:1
H	8/3	2:.75	3:1
P	8/3	2:.75	3:1
A	15/6	3.75:1.5	3:1
R	22/11	5.5:2.7	2:1

Phenotype ratio survey:

T: One hundred and thirty-two people were used in the evaluation of tall:short. Extending from the present generation (born after 1970) back to the 9th generation (born after 1875), the ratio of tall to short was found to be 95 tall:37 short, or a ratio of 3:1. However, while tallness was a dominant phenotype, it was predominantly found in the paternal side extending back from the present generation and shortness was the dominant phenotype extending back from the maternal side of the (writer's) family tree.

D/L/B: One hundred and fifty-three people were used in the evaluation of dark/light hair and baldness. Extending from the present generation back to the 9th generation, the ratio of dark hair to light hair was 115:38, or a ratio of 3:1. However the baldness phenotype appeared to be quite recessive in all families and

showed a ratio of 153:8 or 16:1.

M: Fourteen people in two families in the 12th and 13th generation were observed to determine middigital hair. Eleven persons were found to have middigital hair and three were found not to have it for a ratio of 3:1.

O: When determining the longevity ratio, several factors were taken into consideration. First, not every one in the family tree is dead yet, which narrowed the field considerably. Secondly, not all of the birth and death dates in the genealogical history are complete or unknown. Thirdly, only those persons who died of natural causes (old-age, childbirth, death within one year of the death of a spouse, etc.) were used. Those persons who died at birth, by accident, murder or in wars were not included. Fifty-five people fell into the research category of 75-115 years of age at time of death compared to 50-75 years of age at time of death. The results, 44:11, demonstrate a longevity ratio of 3:1. It was found that longevity was a dominant phenotype on paternal and maternal sides of the family tree in every family looked at.

H: Haemophilia was found to occur only on the maternal side of the writer's immediate family. Of the 8 people (males contract the disease, females are the carriers) in the maternal section of the family tree, only three have haemophilia for a ratio of 3:1. All three are living. The haemophilia has been traced back to the Royal Family of Spain who were directly related to Queen Victoria (Raven, 1988).

P: Premature greying was looked at in persons who had not yet reached an age when hair greys naturally due to the onset of old age. Photographs were used for this observation and no matter what the age of the person observed, only photographs taken of them between the ages of 17 and 27 were considered. Out of the 11 persons observed, premature greying was found in 3 members of one family starting with a grandmother, skipping a generation and reappearing in two first cousins (1 male & 1 female) for a ratio of 3:1.

A: When arthritis was considered for this study, the actual disease of rheumatoid arthritis was not looked at but the arthritic phenotype showing up in the birth of a child whose mother contracted German measles (rubella) while pregnant. The resultant birth abnormalities (Walter, 1977) and malformations are similar to rheumatoid arthritis and ankylosing spodylitis (Walter, 1977). Of the 24 persons observed, 18 did not suffer from arthritis but 6 did. This is a ratio of 3:1. Of the 6 who actually had rheumatoid arthritis, only 1 person (10th generation) had no arthritis at all but had the phenotype of severe rheumatoid arthritis in his hands, arms, and feet. If we consider this person as an F1 then any genetic inherited trait will not show up in his (F2) offspring but will possibly show up as a recessive phenotype in the F3 generation, which it did, in his grandson. It has been shown that arthritis or its symptoms showed up in a 3:1 ratio within the chosen subjects of the study. It has also been shown that within the 6 arthritic persons, the appearance of arthritis has shown up in one person for a ratio of 6:1, but because of the F2 recessive gene it caused it to reappear two generations later, thus bringing about a ratio of 6:2 or 3:1.

R: This particular phenotype, right semi-cauliflower ear did not follow the Mendelian ratio. It was a very dominant characteristic found in only one family and occurred in every generation looked at (through photographs and personal observations, Scarborough, 1991) and was found to have a ratio of 2:1 (out of 22 observed). It was found only in the males and occurred in fathers and brothers and related nephews etc.

CONCLUSION

The patterns of inheritance through the phenotype expression in one family was discussed. The application of the dominant to recessive ratio of 3:1, developed by Gregor Mendel, was discussed. When related to the patterns of inheritance of the family tree researched it was found that the Mendelian ratio was

in fact correct in most of the phenotypes discussed.

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