SELECTIVE LIMITATION OF SIBSHIP SIZE

BY

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The belief that a morbid condition is attributable to their genes may lead parents to limit the size of the sibship in two ways. If the belief precedes the birth of an affected individual (A), the parents may decline to take the risk of producing another. We may then say that the criterion of termination is \( A = 1 \). If they have no such preconception, the birth of a second affected sib may lead them to the same conclusion with the same result. We may then say that the criterion of termination is \( A = 2 \). Such contingencies raise the question: does the decision to terminate the sibship affect the expected proportion of affected individuals in an otherwise random selection of fraternities?

The issue so stated arises frequently in familial studies; and the answer is not so simple as it might appear to be. To clarify it, we shall initially postulate a fixed target value of \( s \), the size the sibship would attain in the absence of any indication relevant to the criterion of termination. This assumption implies the existence of some fraternities which consist of \( s \) members, none of them affected. If \( p = (1 - q) \) be the probability that an individual of given parentage will be affected, the probability of this occurrence is \( q^s \); and \( p \) is also the expected proportion of affected sibs in a complete pool of \( s \)-fold fraternities chosen randomly.

We may hope to obtain a complete pool in this sense, if the criterion of ascertainment is the phenotype of one or both parents, as when the relevant morbid condition is hereditary in the sense formerly current in medical literature. The problem is then on all fours with the issue: what would happen if all parents terminated the sibship on the arrival of a boy? When our concern is with so-called familial conditions, the method of ascertainment excludes fraternities containing no affected members.

The expected proportion of affected sibs in a pool of \( a \)-fold fraternities, otherwise chosen at random, will then be greater than \( p \), being in fact

\[ p_f = \frac{p}{1 - q^a} \quad \ldots \ldots (i) \]

Thus each case (\( A = 1 \) and \( A = 2 \)) distinguished above invites examination on the alternative assumptions that ascertainment is:

(I) complete;

(II) partial in the sense that all recorded sibships contain at least one affected member.

I. ASCERTAINMENT COMPLETE.—If the criterion of termination is \( A = 1 \), the probability \( (pq^a - 1) \) that a sibship will contain one affected sib, preceded by \( (a - 1) \) not affected is the probability that a completed family containing at least one affected will consist of \( a \) sibs.

The total number of sibs is \( a \) times the number of affected sibs in such a sibship. There will also be a sibship which terminates without producing an affected member. If \( s \) is the maximum size of the sibship, the probability of this occurrence is \( q^s \). If the method of ascertainment is through the parents, we shall include such sibships and the expected proportion of affected sibs is:

\[ \Gamma_0 = \frac{\sum_{a=1}^{a=s} p(aq^a - 1)}{sq^a + \sum_{a=1}^{a=s} apq^{a-1}} \quad \ldots \ldots (ii) \]

When the criterion of termination is \( A = 2 \), the completed \( a \)-fold family containing two affected sibs is one in which a sequence of \( (a - 1) \) containing only one affected sib precedes an affected sib of birth rank \( a \). The probability of this occurrence is \( (a - 1) p^2q^{a-2} \). Among such sibships the ratio of all sibs to those affected is \( a : 2 \).

If \( s \) is the maximum size of the family, there will be sibships containing only one affected sib and sibships containing none. The probabilities of these occurrences are respectively \( sq^{a-1} \) and \( q^a \). The ratio of total affected in the sibships with only one affected member is \( s : 1 \). If ascertainment is complete we therefore derive:

\[ p_0 = \frac{sq^a - 1}{sq^a + sq^{a-1} + \sum_{a=1}^{a=s} a(a - 1) p^2q^{a-2}} \quad \ldots \ldots (iii) \]

To evaluate \( (ii) \) we require the following summation formulae:

\[ \sum_{a=1}^{a=s} pq^{a-1} = 1 - q^s, \]

and

\[ \sum_{a=1}^{a=s} apq^{a-1} = p - 1 (1 - q^a) - sq^s \quad \ldots \ldots (iv) \]
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Whence, from (ii), we obtain for complete ascertainment when the criterion of termination is $A = 1, p_o = p$. To evaluate (iii) we require the additional identity:

$$\sum_{a=2}^{a=s} a(a-1)p^2q^{a-2} = \frac{2(1-q^a)}{p} - s^2pq^{s-1} - sq^{s-1}(1+q).$$

Whence we obtain again $p_o = p$, when $A = 2$. Thus the decision to terminate the sibship when $A = 1$ or $A = 2$ does not lead to a biased estimate of $p$, if ascertainment is complete. It is easy to discern the pattern common to the two cases specified by (ii) and (iii), hence to show that the result stated holds good for $A = 3$, etc. Since the target value $s$ does not enter into the outcome, the argument holds good for any value $s$ may have; and the arbitrariness of the initial assumption that $s$ has a fixed value does not invalidate it.

II. ASCERTAINMENT PARTIAL.—For a sibship of $a$ members (i) prescribes $ap(1-q^{a-1})$ as the expected number of affected sibs in fraternities containing at least one such. If $N_a$ is the observed number of affected in $n_a$ $a$-fold sibships which terminate in the production of an affected sib, and $E_a$ is the expected number calculated on the assumption that the production of an affected sib does not influence the decision of the parents to terminate the sibship, then

$$N_a = \frac{1 - q^a}{E_a}.$$

For $a = 1, 2, 3$ etc. the values of this ratio are:

$$\frac{1}{2}, \frac{1 + q}{2}, \frac{1 + q + q^2}{3}, \ldots \ldots \text{etc.}$$

Since we assume that $q < 1$, all the terms of this series are less than unity if $a > 2$. Thus $N_a < E_a$ for all values of $a > 2$. In short, the effect of deliberate termination of the sibship upon the arrival of an affected sib must make the number of affected less than the expectation computed in accordance with (i).

When $A = 1$, every sibship in a pool of fraternities containing at least one affected member will conform to the criterion of termination; but $s$-fold fraternities may contain two or one when $A = 2$, since the sibship may reach its limiting size without the birth of a second affected member. If $n_a$ is the number of fraternities of $a$ sibs, the expected proportion of affected sibs computed in accordance with (i) will in any case be:

$$p_e = \frac{\sum_{a=2}^{a=s} n_a \cdot a \cdot p^2 q^{a-2}}{\sum_{a=2}^{a=s} a \cdot n_a}.$$

To see what value $p_e$ would have when we apply (i) to a pool of sibships selectively limited in accordance with the criterion of termination $A = 2$, we have to weight

(a) each $a$-fold sibship for values of $a$ from 2 to $(s - 1)$ in conformity with the prescribed distribution of family size, $n_a$ being thus proportional to $(a - 1)p^2q^{a-2}$;

(b) $s$-fold families with due regard to those which terminate without producing more than one affected sib, so that $n_s$ is proportional to $(s - 1)p^2q^{s-2} + spq^{s-1}$.

We should thus obtain:

$$s^2p^2q^{s-1}(1-q^a)^{-1} + \sum_{a=2}^{a=s} a(a-1)p^3q^{a-2}(1-q^a)^{-1}$$

$$p_e = \frac{\sum_{a=2}^{a=s} a(a-1)p^3q^{a-2}}{s^2pq^{s-1} + \sum_{a=2}^{a=s} a(a-1)p^2q^{a-2}}. \ldots \ldots \text{(vi)}$$

The corresponding value of $p_o$ obtainable from (iii) by elimination of one term in the denominator is:

$$spq^{s-1} + \sum_{a=2}^{a=s} 2(a-1)p^2q^{a-2}$$

$$p_o = \frac{\sum_{a=2}^{a=s} a(a-1)p^2q^{a-2}}{s^2pq^{s-1} + \sum_{a=2}^{a=s} a(a-1)p^2q^{a-2}}. \ldots \ldots \text{(vii)}$$

The case $p = \frac{1}{2}$ is of minor interest in this context, since it implies that one parent is affected. Thus the appearance of a single affected offspring would suggest what in common parlance is a family taint. Thus the considerations likely to lead to $A = 2$ as the criterion of termination when neither parent is affected ($p = \frac{1}{4}$) would dictate $A = 1$ as the criterion when in the alternative situation. We need only therefore examine the values of $p_e$ and $p_o$ as exhibited in (vi) and (vii) for different values of $s$, the assumed absolute limit to fraternity size:

<table>
<thead>
<tr>
<th>$s$</th>
<th>$p_o$</th>
<th>$p_e$</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>0.571</td>
<td>0.571</td>
</tr>
<tr>
<td>3</td>
<td>0.439</td>
<td>0.443</td>
</tr>
<tr>
<td>4</td>
<td>0.376</td>
<td>0.383</td>
</tr>
<tr>
<td>5</td>
<td>0.339</td>
<td>0.350</td>
</tr>
</tbody>
</table>

Thus there is a bias in the direction we might surmise from a consideration of the case $A = 1$; but it is trivial in situations involving family limitation as commonly practised in contemporary civilization.
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