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CASE REPORT

Psychomotor developmental delay and epilepsy in an offspring of father–daughter incest: quantification of the causality probability

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Abstract A 20-year-old offspring of father–daughter incest, who has been suffering from serious psychomotoric health problems since early childhood, is seeking financial compensation under the German federal act of victim indemnification. For her appeal to be valid, the probability X that the incest was causal for her disorder must exceed 50%. Based upon the available medical records, we show that this is indeed the case and that X is even likely to exceed 65%, thereby rendering the victim's claim scientifically and legally justified.

Keywords Incest · Causality · Autozygosity · Epilepsy · Syndrome

Introduction

A regional pension office in Germany ("Landesversorgungsamt") requested an expert opinion on the probability that the severe health problems experienced by a 20-year-old woman (AB) were causally attributable to the fact that she was an offspring of father–daughter incest. If the probability was larger than 50%, the woman would be entitled to a life-long pension on the basis of the federal act of victim indemnification ("Opferentschädigungs-Gesetz"). However, the legal custodian of AB had denied permission to any additional physical or genetic examinations so that the expert opinion had to be based on the available medical records.

Since early childhood, AB had suffered from severe psychomotor developmental delay, therapy-resistant epilepsy with myoclonic and tonic seizures, and grand-mal attacks. Developmental delay and seizures were reported to have commenced at approximately 4 months of age. Hearing impairment was first noted and a partial optic atrophy suspected at 15 months old. Microcephaly was first documented when AB was 2 years old. Computer tomography revealed symmetric cerebral atrophy with ventricular enlargement both at 9 months and at 2 years 8 months of age. When she was 10 years old, AB was suspected of having Angelman syndrome, but this diagnosis could not be confirmed. The clinical picture of AB is also reminiscent of autosomal-recessive infantile epilepsy syndrome [1], but a direct, molecular genetic test for this disorder could not be performed owing to the abovementioned disapproval of closer examination by the legal custodian. Thus, no definite diagnosis of AB's apparently syndromic disorder has ever been made.

Formally, the probability P that a syndromic disorder like that of AB arises in an offspring of father–daughter incest can be written as

$$P = P_1 + P_2 + P_3 \quad (1)$$

where P_1 equals the probability that the at-risk individual is autozygous for at least one mutation which in the homozygous state causes the disorder in question (henceforth referred to a 'causally autozygous'), P_2 summarizes other incest-specific factors not included in P_1, and P_3 is the base-line risk for the disease in the general population.

If an incest offspring is causally autozygous, the logical implication is that the incest was indeed "causative" of their disease in the sense of the German victim indemnification

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act. Therefore, the sought-for probability X is at least as large as the probability W that an incest offspring is causally autozygous. This probability can be calculated from Bayes' theorem as

$$W = \frac{P_1}{P} \tag{2}$$

In other words, any sensible estimate of W automatically provides a lower limit for X.

According to the scientific literature, the total disease probability P amounts to 0.30 [2] whereas P_3 equals 0.05 [3]. Furthermore, the unknown probability P_1 can be extrapolated from information on the offspring of first cousin marriages. For such children, the probability of causal autozygosity equals ¼·P_1, rather than P_1, and the total syndromic disease risk reportedly lies between 8% [2] and 10% [4]. Taking averages yields

$$\frac{1}{4}P_1 + P_3 = 0.09 \tag{3}$$

so that P_1 approximately equals $4\times(0.09-0.05)=0.16$.

Entering the above estimates of P and P_1 into formula (2) yields $W=0.16/0.30=0.533$, or 53.3%. However, as pointed out above, this value is likely to represent a lower limit to the true causality probability X. Some of the risk factors subsumed in P_2 will be direct consequences of the father–daughter incest, including an inherited and psychosocial burden to both the mother and the child that would not be properly accounted for by extrapolating P_1 from first cousin marriages. Even if only half of the value of P_2 (i.e., of $0.030-0.16-0.05=0.09$) were attributable to such factors, and would thus be added to the numerator in formula (2), then $W=0.205/0.30=0.683$, or 68.3%.

Although the scarceness of data did not allow an exact enumeration of the probability X that AB's disease was caused by her stemming from father–daughter incest, the available information justifies the conclusion that X is certainly larger than 50%, and more realistically exceeds 65%. According to German law and jurisdiction, AB is thus entitled to financial compensation from public resources. A decision about her appeal for victim indemnification is currently pending.

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