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Synaesthesia: prevalence and familiality

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Abstract. Synaesthesia is a condition in which a mixing of the senses occurs; for example, sounds trigger the experience of colour. Previous reports suggest this may be familial, but no systematic studies exist. In addition, there are no reliable prevalence or sex-ratio figures for the condition, which is essential for establishing if the reported sex ratio (female bias) is reliable, and if this implicates a sex-linked genetic mechanism. Two independent population studies were conducted in the city of Cambridge, England (studies 1 and 2 here), as necessary background to the family genetic study of synaesthesia (study 3). Studies 1 and 2 arrived at an almost identical prevalence rate for synaesthesia: approximately 1 case in 2000. The sex ratio found was 6:1 (female:male). A third of cases also reported familial aggregation. In study 3 six families were examined, and first-degree relatives were tested for genuineness of the condition. All six families were indeed multiplex for synaesthesia. Alternative modes of inheritance are discussed.

1 Introduction

Synaesthesia occurs when an individual experiences a sensation in one sensory modality triggered involuntarily and automatically by a sensation in a different sensory modality (Motluk 1994). The most common form appears to be seeing colours when hearing sounds. Typically, in 'coloured hearing', the person sees a different colour when hearing a different sound, but in a highly consistent fashion. For example, when speech is heard, each word heard triggers a different colour, and over time the same word always triggers the same colour. Synaesthesia appears to be consistent across the individual's whole lifetime, and is present from as early in childhood as the individual can recall. Synaesthesia can occur between any two sensory modalities, though in practice some combinations are more common than others.

One hundred years ago, synaesthesia was a topic of considerable scientific interest (Binet 1893; Myers 1911, 1914). By the 1940s, the topic had virtually vanished from science, for two reasons: introspection had become an unrespectable method of data collection in experimental psychology, and there appeared to be no objective way of validating that synaesthesia was actually occurring, over and above the self-report data from the subjects themselves.

In a study of a single case (EP) Baron-Cohen et al (1987) aimed to overcome the problem of self-report data being unreliable. They showed that when EP was given a long word list and was asked to describe the colours triggered by each word in the list, she gave very detailed descriptions (for example, the word 'Maria' triggered a dark purple colour, with a shiny texture, and with speckled spinach-green at the edges). When retested on the same word list without any prior warning one year later, she was 100% consistent in the colours she described for each word. In contrast a normal control subject, matched for intelligence and memory, who was asked to associate colours with words in the same word list, and who had the advantage that she was warned that she would be retested after 2 weeks (and so to attempt to use mnemonics), was only 17% consistent. Since a memory strategy could not plausibly account for

such performance, it was concluded in the study that synaesthesia was a genuine phenomenon. This finding was replicated on a larger group of synaesthetes in a later study (Baron-Cohen et al, 1993).

Positron emission tomography has been used to investigate if there is a neural basis to synaesthesia (Paulesu et al 1995). Six synaesthetic women were compared with six matched controls. Results showed that when hearing words whilst blindfolded, the group of synaesthetes showed abnormal activation in some areas of visual association cortex (the posterior inferior temporal cortex and the parieto-occipital junctions), relative to controls. A recent study of a single synaesthete by means of functional magnetic resonance imaging showed a clear result of visual-cortex activation when words were heard, in contrast to a normal control (Gray et al 1997). This is clearly an abnormal finding, and suggests that visual imagery is occurring when words are heard. One possibility is that this is due to there being abnormal neuronal connectivity between auditory and visual cortical areas in synaesthesia.

The neuroimaging studies suggest synaesthesia has a biological basis. Further evidence for this comes from studies showing that synaesthesia can be induced in normal individuals by hallucinogenic drugs such as LSD and mescaline (Cytowic 1994; Motluk 1994). It should be stressed however that the subjects in the studies cited above and reported below have synaesthesia naturally—they are not taking any form of drug.

A further line of evidence that synaesthesia has a biological basis is the suggestion that it may be genetic. Baron-Cohen et al (1993) reported that of 565 individuals who claimed to have synaesthesia, more than 95% were female, suggesting the condition may be sex-linked. Cytowic's (1994) studies also suggest a predominance of females in samples of synaesthetes. Baron-Cohen et al (1993) carried out a questionnaire survey of these 565 individuals with synaesthesia and asked about other family relatives. In 132 of these cases (23.4%), a first-degree relative was also reported to have the condition, and in all cases the affected relatives were female. However, this survey has the weakness of being a postal questionnaire; what is needed is a test of relatives to validate the apparent familial pattern to the condition. We report the first such test (study 3).

Currently, there is no way of estimating how common synaesthesia is. The only reported statistic is from Cytowic (1994), who states that it is 1 per 100 000. However, he admits that this is a guess based on how many cases he has encountered in the USA (personal communication). Motluk (1994) suggests it is 1 in 25 000, but again this was not based on any study. In this paper, this claim is tested by carrying out the first population studies of synaesthesia (studies 1 and 2 below).[1] This is necessary background to the family genetic study (study 3).

2 Methods

In Study 1 (the first prevalence study), an advert was placed in the *Cambridge Evening News*, asking people who experienced synaesthesia to contact the research group. The readership was 44 000, drawn almost exclusively from the town rather than the university.[2] In study 2 (the second prevalence study), the identical advert was placed in the Cambridge University student magazine *Varsity*, which has a readership of 11 000.

[1] Note that the method used is not strictly epidemiological. This is because a priori there is no way of knowing what size population to screen, given the previously conflicting reports. Instead, as a first approach to this problem, a method was used that could establish the minimum prevalence of synaesthesia.

[2] The wording of the advert was as follows: "Research into Synaesthesia: Some people experience a mixing of the senses. This is known as Synaesthesia. For example, whenever they hear sounds, they automatically see colours. For other people, a different sense (touch, taste, or smell) might trigger a visual or auditory experience. We are trying to find out how common this is, and would like to hear from people who think they may have Synaesthesia." This wording was designed to avoid biasing ascertainment of any particular kind of synaesthesia.

This population is drawn almost exclusively from the university, rather than the town. The two prevalence studies were therefore of nonoverlapping populations. Note also that whereas in study 1 a population of mixed educational levels was screened, in study 2 the population was of high educational level.

Responders to both studies were contacted by two of the authors (FSL and LB) and tested for genuineness, and their kind of synaesthesia was categorised. This was done by inviting the person into the laboratory in the University. In study 3 (the family study), six families were selected from the International Synaesthesia Association's database in London.[3] The six families were selected from these apparently familial cases, on the basis that they lived in the South of England (for convenience in terms of testing them). In all other respects, their selection was random. Of the six families examined, each first-degree family member was tested individually in the laboratory in Cambridge. A family history was also taken, by interview.

The test of genuineness for coloured-word synaesthesia (CWS) was used to verify cases in all three studies. This was a modified form of that used in Baron-Cohen et al (1987), involving seventy items (words, letters, numbers, and nonsense words). To score responses, a colour chart of 309 graded and numbered colours was used. The experimenter read aloud each of the seventy auditory stimuli, and for each stimulus the subject was asked to point to the nearest match on the colour chart that corresponded to the colour they saw in their mind. Subjects were retested after an interval of at least one hour, in order to score consistency. Scoring of consistency was carried out by two independent judges, and only in those cases where there was independent agreement was the item scored as consistent.

3 Results

In study 1, twenty-eight people responded to the advert, all of whom were tested. None were students. Eighteen met criteria for genuineness on CWS, and four for coloured-music synaesthesia (CMS). Two cases simply had colour associations for days of the week. Since this was considered not to be synaesthesia, they were not tested further. Four others claimed to have synaesthesia, but instead simply had a strong interest in colour or art. Thus, twenty-two cases of genuine synaesthesia were found in the population of 44 000, corresponding to a prevalence of 1 per 2000. The sex ratio of the twenty-two synaesthetes was nineteen females to three males, corresponding to a sex ratio of 6.3:1 (female:male). Handedness was 15:1 (right:left). Mean consistency of the group was 97.58% (SD 4.97%). A control group of eleven normal subjects, matched on age, sex, and educational level, had a mean consistency of 19.09% (SD 15.78%). This group difference is highly significant ($t_{22} = 17.023$, $p < 0.0001$).

Of the cases of CWS, all had coloured-letter synaesthesia [called 'chromatic-graphemic' synaesthesia in Baron-Cohen et al's (1993) study], but in all of these cases whole words triggered colours, and all individuals also had experiences of words for which the colour did not match the colours of the individual letters. Note also that 40.9% of the subjects with CWS also claimed to have CMS (though this was not systematically assessed), and 72.7% of the subjects with CWS also had coloured numbers. 13.6% of those with CWS also reported that they saw colours triggered by other auditory stimuli (nonwords, nonmusic). Last, seven subjects (or 33% of the sample) reported having a relative who also had synaesthesia.

In study 2, four people responded, all of whom were students, all of whom were tested, and all of whom met criteria for genuineness. Their mean overall consistency was 96.5%. This gives a prevalence rate of 4 per 11 000, or 1 per 2500. The sex ratio

[3] This is a register of 565 synaesthetes (at the time of the study). Approximately 26.4% of these claim to have other family members who also have synaesthesia, as established by questionnaire (see section 1).

was 3:1 (female:male), and all four subjects were right-handed. All four had CWS. One case out of four had a relative who also had synaesthesia. Thus in study 2 a very similar prevalence rate of synaesthesia to that was found in study 1, suggesting this is likely to be a true estimate, and again confirms the sex difference (female bias). A similar proportion (25%) to that found in study 1 also reported familial aggregation.

In study 3, the family pedigrees collected for each family are shown in the appendix. This confirms that all six families are multiplex for synaesthesia.[4] The familial prevalence rate of synaesthesia is 48.6% among first-degree relatives, whilst the population prevalence rate is 0.05%. This strongly demonstrates familiality.

4 Discussion

Studies 1 and 2 establish that the minimum population-prevalence rate of synaesthesia is 1 in 2000. Study 3 demonstrates that synaesthesia is familial. This is unlikely to be the result of learning, for the following reasons. (i) On average, each family only had four letters of the alphabet with common colours, which suggests imitation and cultural learning is unlikely to be the cause. (ii) In two families, the children had different types of synaesthesia from their parents, which again argues against a learning explanation. (iii) In these families, vowels were the same colours as have been reported in previous studies, with data from widely different times in history and from different cultures (Baron-Cohen et al 1993). It is highly unlikely that the same coloured-alphabet teaching method would have been used. This therefore implicates a neural basis to these connections. (iv) In these families (as in previous studies), a run of letters in the alphabet often triggers shades of similar colours (eg L, M, and N might be all shades of blue). It is hard to imagine alphabetic teaching methods being designed using such a principle, since if anything such a design might only confuse child learners.

The results of study 3 are instead consistent with a genetic cause to synaesthesia. But this raises the question as to its mode of inheritance. Here we consider three different possible models.

4.1 *Autosomal recessive*:

Bailey and Johnson (1997) suggest this mode of inheritance is unlikely, for three reasons: (a) the chance of an unaffected homozygote having married an unaffected heterozygote is low; (b) it would lead to a predicted 50% affected offspring of both sexes, which is not observed here; and (c) the vertical transmission through several generations also rules out autosomal recessive inheritance, and also makes polygenic inheritance unlikely. We can therefore dismiss this mode, and consider two other options.

4.2 *Autosomal dominant, with sex limitation*:

Cytowic (1989) suggested, from the familial cases he observed, that synaesthesia can be passed down either the paternal or the maternal line, to offspring of either sex, with no skipped generations. This would fit an autosomal dominant mode of inheritance, but would not make sense of the sex ratio observed unless it was with sex limitation (reduced expression in males). At the present time, this is not ruled out.

4.3 *Sex-linked dominant, with lethality*:

Bailey and Johnson (1997) concur with us that the data best fit a sex-linked (X-linked) dominant mode of inheritance, with lethality, for several reasons: (a) there is a high rate of transmission from affected males to affected daughters (5/5 meioses); (b) the proportion of affected offspring from maternal transmissions is a little over 50%, as expected under this mode of inheritance; (c) the strong sex difference in all

[4] Two children were only 61% consistent—both aged 14 years—but since this is significantly higher than either chance levels or control-group baseline levels, they are included here. It may be that consistent reporting among children with synaesthesia is less clear than in adults.

studies to date (as high as 6:1, female:male) does not fit with a simple sex-linked dominant condition. It might lead to a bias of as high as 3:1 (female:male), since in an X-linked dominant condition affected heterozygous females will transmit the trait to half their offspring of both sexes, but males transmit it to all female offspring and to no male offspring. But to account for a ratio of >3:1, as appears to be the case with synaesthesia[5], one would need to posit X-linked inheritance with lethality in hemizygous males. Bailey and Johnson clarify that the lethality aspect (not the synaesthesia aspect) would be recessive, acting only in males, where no normal allele is present. This would lead to the prediction that among the offspring of affected mothers, the overall sex ratio would be heavily biased towards females, since 50% of the male foetuses should die in utero. The figures for the six families reported in study 3 are eighteen female to nine male offspring, which exactly matches this prediction. Bailey and Johnson add that a further prediction under this model would be an increase in miscarriages amongst women with synaesthesia. At the present time there is no relevant miscarriage data with which to test this prediction. Last (d), there should be no male-to-male transmission, since male offspring get their X chromosome from their mother. Affected males must therefore have had an affected mother (irrespective of whether the father was affected). It is noteworthy that the novelist Vladimir Nabokov (a male synaesthete) had two synaesthetic parents, so this is not inconsistent with this model. None of these predictions are disconfirmed by the data presented here. Of course, further research will be needed to test this model, with larger samples.

4.4 *Risk rates for relatives*

Irrespective of the genetic mechanism, the results show that, if another person in the family also has synaesthesia, then the risk for a daughter of an affected proband of having synaesthesia is 72.2% (based on there being eight out of eleven affected daughters in the six affected families studied), and the risk for a son of an affected proband having synaesthesia is 25% (based on there being one out of four affected sons in the six affected families studied). The risk for sisters of affected probands is 60% (three of five sisters were affected, in the six affected families), and the risk for brothers was 16.6% (based on one in six being affected).[6]

The idea that a single gene might control complex mental phenomena may seem remarkable. It is noteworthy that if this model of synaesthesia is correct, it is not an isolated case in nature. Three other examples can be found within psychiatric genetics. First, Gilles de la Tourette Syndrome shows all the hallmarks of being a single-gene disorder (Robertson 1994). In this condition, children and adults experience involuntary movements (motor tics), involuntary vocalisation (vocal tics), and involuntary thoughts (intrusive obsessions). Secondly, X-linked mental retardation syndromes, of which there are several, have the expression of impaired cognitive processing (Neri et al, 1994). Last, colour-blindness (like synaesthesia) results from a single gene and changes perceptual experience (Vogel and Motulsky 1992).

If synaesthesia is caused by a genetic mutation, is there any evidence that synaesthesia is maladaptive? Clearly, not all mutations are maladaptive, but this is one possibility. Recently, in the television programme *Horizon* (BBC2, 13 December 1994) the case of JR was described, a 45-year-old female music teacher who had coloured hearing in *both* directions; that is, she saw colours when she heard sounds, and also heard sounds

[5] It may of course be that the true sex ratio in synaesthesia is not as high as 6:1, and is more like 2:1 or 3:1. We will need more truly epidemiological studies to check this possibility.

[6] Note that since in studies 1 and 2 the rate of familial synaesthesia found was approximately 30%, first-degree-relative risk for daughters can be estimated as $0.3 \times 0.72 = 0.22$, and the first-degree-relative risk for sons can be estimated as $0.3 \times 0.25 = 0.075$. Such risk rates are significantly higher than the population rate.

when she saw colours. We have tested JR extensively, and confirm that she shows high consistency in both directions. As she looks at a visual scene, each colour triggers a different musical note, and as she hears different sounds (speech or other sounds), each of these triggers its own colour. Not surprisingly, JR suffers considerable perceptual interference and stress as a result of her form of synaesthesia, and copes with this only by leading a relatively restricted lifestyle, avoiding noisy environments or 'loud' colours in the environment. It may be then, that whilst milder cases of synaesthesia carry no selective disadvantage, certain forms of synaesthesia are indeed maladaptive. One implication of this is that the *modularity* characteristic of the senses in nonsynaesthetes may have been selected for because it leads to more-efficient information processing. The individual can be sure of whether he or she heard something or saw something, and is not confused over this. Whether normal perception is highly modular, as Fodor (1983) suggests, and synaesthesia is an example of "modularity breakdown" (Baron-Cohen et al 1993; Baron-Cohen 1996) is an issue which merits further testing.

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APPENDIX

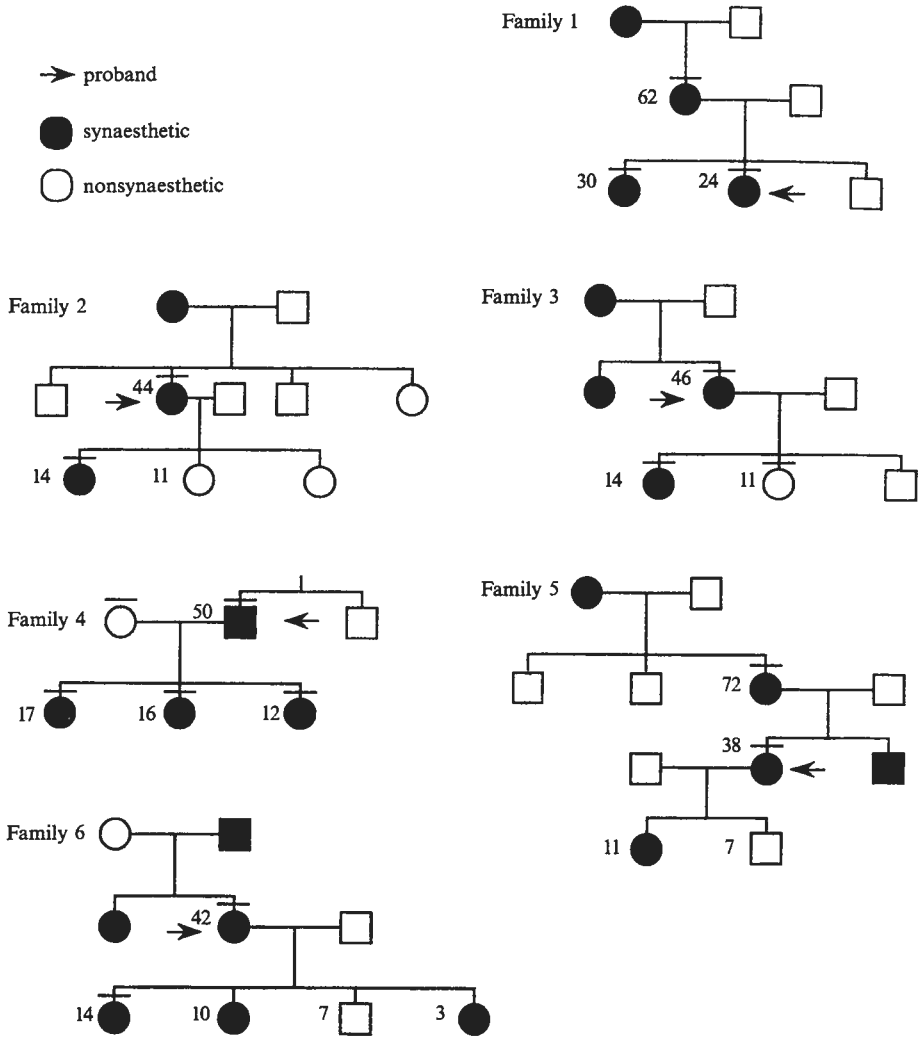

Figure A1. Family pedigrees from study 3. Squares represent males, circles females. Horizontal lines indicate the individual was actually tested, rather than information being derived solely from report. Numbers indicate the individual's age.