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# WHY DID THE SAVANT SYNDROME NOT SPREAD IN THE POPULATION? A PSYCHIATRIC EXAMPLE OF A DEVELOPMENTAL CONSTRAINT<sup>4</sup>

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### Abstract

*A developmental constraint is a mechanism that limits the possibility of a phenotype to evolve. There is growing evidence for the existence of developmental constraints in the biological literature. We hypothesize that a developmental constraint prevents the savant syndrome, despite its positive aspects, from spreading in the population. Here, the developmental constraint is the result of the high interactivity among body parts in an early stage in embryological development, namely early organogenesis or the phylotypic stage. The interactivity during this stage involves all components of the embryo, and as a result mutations that affect one part of the embryo also affect other parts. We hypothesize that a mutation, which gives rise to the development of the positive aspects of the savant syndrome (e.g., an impressive memory capacity), will virtually always have a deleterious effect on the development of other phenotypic traits, (e.g., resulting in autism and/or impaired motor coordination). Thus, our hypothesis states that the savant syndrome cannot spread in the population, because of this developmental constraint. The finding that children with savant syndrome often have autism and physical anomalies, which are known to be established during early organogenesis, supports our hypothesis.*

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A developmental constraint is a developmental mechanism or process that limits the ability of a phenotype to evolve, or biases it along certain pathways (Maynard-Smith et al., 1985; Amundson, 1994, 2005; Beldade et al., 2002; Schwenk and Wagner, 2003). In the biological literature, the evidence for the existence of developmental constraints is growing (Amundson, 2005). In the psychological literature, the concept of constraint is often considered, but is generally accorded a different meaning. In this chapter, we first explain the different conceptualizations of the term constraint in fields of psychology and biology. Then we discuss biological examples to illustrate the concept of developmental constraint, and we present what we contend to be an instance of a developmental constraint in the psychiatric context. Specifically, we hypothesize that it is a developmental constraint that prevents the positive aspects of the savant syndrome to become established in the population through the process of natural selection.

### **Conceptualizations of the concept of constraint**

In the psychological literature, the term constraint is generally used in the context of learning. Keil (1981), in a now classic article on constraints on knowledge and cognitive development, defined constraints as ‘formal restrictions that limit the class of logically possible knowledge structures that can normally be used in a given cognitive domain’ (p. 198). Although the use of the term *limit* seems to imply that constraints hinder the child in its learning, it is rather believed that constraints facilitate learning. This is because constraints guide the learner towards a non-random selection of logically possible characterizations of information (see also Keil, 1990). For an instance of such a constraint, Keil (1981) refers to Chomsky’s (1965) work on children’s language acquisition. Here constraints are thought to be necessary to enable children to develop language as quickly and easily as they do. Moreover, constraints have been called *learning enablers* (Gelman & Williams, 1998). Thus, as was put forward by Bjorklund and Pellegrini (2002), constraints help children to make sense of certain types of information, and are likely to be the outcome of natural selection.

In the field of biology, the concept of developmental constraint is accorded a different meaning. Here, developmental constraints are related to the relative imperviousness of existing developing structures to modifying or reorganizing influences (Raff, 1996). Although many different biological variants are imaginable, only a limited number of variants seem to arise. From an

evolutionary perspective, this is remarkable, because an abundance of variation may be expected, as it is variation upon which natural selection acts.

### **Biological examples of developmental constraints**

A biological example of a developmental constraint is the fact that we have five, and not more, fingers (Galis, van Alphen, & Metz, 2001). This is remarkable, because a common anomaly at birth is the presence of an extra finger or toe (e.g., in humans 0.1-0.2%; Castilla et al., 1996). Thus, there is variation in the number of digits, but extra digits have never become a stable pattern. An easy conclusion would be that extra digits do not have a selective advantage. However, extra digits may well be advantageous for climbing, digging, or swimming, and they have no apparent disadvantages. In fact, many animals have evolved extra digital structures, but these are never 'true' digits, but rather are modified wrist bones or extra phalanges (Galis et al., 2001; for example, the 'panda's thumb', see Gould, 1980). Thus there appears to be a developmental constraint on digit number that cannot easily be explained in terms of natural selection.

Galis et al. (2001) hypothesized that this developmental constraint is due the fact that limb patterning occurs very early in embryonic development, namely during early organogenesis (sometimes referred to as the phylotypic stage). This stage is characterized by intense inductive signaling, a process of massive response induction among cells. The result is that genetic mutations in one part of the embryo affect other parts of the embryo (so-called *pleiotropic effects*). If a mutation for an extra digit occurs in this stage, it is very likely that changes in another part of the body also occur, usually with deleterious effect. Infants born with an extra digit often have other congenital abnormalities, such as spina bifida, cleft lip, microcephaly, 13 trisomy syndrome, Meckel syndrome, and Down syndrome (Castilla et al., 1998). Polydactyly is also associated with serious abnormalities in other animals (for references, see Galis et al., 2001). This is probably the reason for the (indirect) selection against extra digits.

There are several other examples of developmental constraints in the biological literature. For instance, there is a developmental constraint that prevents parthenogenesis in mammals, i.e., the production of offspring solely from maternal germ cells (Kono et al., 2004). Without bi-parental parenting there is unequal expression of imprinted genes from maternal and paternal

alleles (Tilghman, 1999). Some genes are differently imprinted in maternal and paternal chromosomes, and both imprintings are necessary for normal development. For example, if chromosome 15q13 misses paternal imprinting, the result is the Prader-Willi syndrome (Butler & Palmer, 1983); if maternal imprinting is missing, the result is Angelman syndrome (Williams et al., 1990).

A third example of a developmental constraint was established in the fruit fly *Drosophila*. Genes that are expressed only during early embryonic development have been duplicated significantly less often during evolution than genes expressed in the later larval stage (Yang & Li, 2004). The duplication of genes is regarded as a major source of evolutionary change, so there appears to be a developmental constraint that prevents modification of early embryonic development. In a study of the worm *C. Elegans*, Yang and Li also found that genes expressed early in development have been duplicated less than those expressed late in development.

### **A psychiatric example of a developmental constraint**

An example of a developmental constraint in psychiatry is related to the savant syndrome. Sufferers of this rare syndrome are often referred to as idiot savants, retarded savants, or autistic savants. They combine a serious disability, often mental retardation and/or a pervasive developmental disorder, such as autism, with a spectacular ability, which stands in stark contrast with the handicap (Treffert, 2000). The ability is highly developed in a narrow range, such as music or art performance, calendar calculating, mathematics, and mechanical or spatial skills, coupled with a phenomenal memory within the area of the skill itself (Miller, 1999; Treffert, 1999, 2000). In some savants, the skills are remarkable only in contrast to the handicap (Treffert calls such savants *talented*). In other savants, the skills would be remarkable even if observed in an otherwise normal person (Treffert calls savants with these skills *prodigious*). The skills themselves would probably be advantageous for many individuals by increasing the survival and reproductive chances. However, because these skills co-occur with a disability, natural selection does not favor these abilities.

We hypothesize that the savant syndrome has a genetic basis, and that the genes involved in the development of this syndrome have pleiotropic effects (i.e., affect more than one phenotypic trait). Furthermore, we hypothesize that the side-effects of this syndrome are unavoidable due to its developmental origin in the highly interactive early organogenesis stage. The exceptional skill

associated with the syndrome is itself a positive pleiotropic effect. However, this positive pleiotropic effect is always accompanied by negative pleiotropic effects, which nullify any selective advantage. The negative pleiotropic effects serve as a developmental constraint; because of this constraint, the savant syndrome cannot spread in the population, despite its positive aspects.

As our hypothesis is based on the supposition that the savant syndrome has a genetic basis, we first must establish this. Nurmi et al. (2003) reported that autism combined with savant syndrome is genetically linked to chromosome 15q11-q13, but Ma et al. (2005) could not replicate this finding. Despite this controversy about the specific genes involved in savant syndrome, there is little reason to doubt that the savant syndrome has a genetic basis. Savant syndrome is linked to autism (Treffert, 2000; Heaton & Wallace, 2004), and the genetic basis of autism is well-established (for a recent review, see Klauck, 2006). For example, Steffenburg et al. (1989) found a concordance rate for autism of 90% in identical twins, compared with 0% in fraternal twins in a Scandinavian sample. Bailey et al. (1995) found a concordance rate of 73% in identical twins, compared to 0% in fraternal twins in a British sample. Based on a large number of family and twin studies, Bailey et al. (1996) estimated the heritability of autism to be at least 90%. Thus, because of the co-occurrence of autism and the savant syndrome, it seems very likely that the savant syndrome also has a genetic basis.

Second, our hypothesis presupposes that the positive aspects of the savant syndrome are accompanied by negative aspects. There is ample support for this hypothesis, as was reviewed by Miller (1999) and Treffert (2000). Characteristics of savants include pervasive developmental disorders, such as autism, convulsions, visual impairment, deafness, Tourette syndrome, epilepsy, schizophrenia, and language disorder or delay. Most savants are mentally retarded, although there are some exceptions (Heavey et al., 1999; Young & Nettlebeck, 1994). All studies on savants indicate that subjects with remarkable abilities in a narrow range suffer disabilities that prevent them from living independently, and, importantly from an evolutionary perspective, prevent them from acquiring a partner and enjoying reproductive success. For example, fifty percent of the savants have autism (Treffert, 2000), a disorder that clearly prevents sufferers from attracting and retaining sexual partners (Larsen & Mouridsen, 1997; Keller & Miller, 2006). Some have argued that autism is not a necessarily a harmful disorder, because people with autism outperform people without autism on numerous perceptual, reasoning, and

comprehension tasks (Gernsbacher et al., 2006). However, as long as people with autism do not benefit from this superiority in attracting sexual partners, the disorder will be harmful from the evolutionary perspective (i.e., selectively disadvantageous). Because of the disabilities of the savant, whether these include autism or other handicaps, the exceptional skill of the savant will not spread in the population. This is not because of selection against the exceptional skill, but because of selection against the accompanying severe disabilities.

Third, our hypothesis posits that the pleiotropic effects originate in the early organogenesis stage, i.e., very early in embryological development. Pleiotropic effects in this stage generally serve as a much stronger developmental constraint than pleiotropic effects in later embryological stages, because that have more pervasive effects (Galis & Metz, 2001). Support for this idea comes from several sources. First, many savants have brain anomalies (for an overview, see Treffert, 2000, pp. 193-198). Most savants with brain damage have left-side abnormalities. Because the brain starts to develop during early organogenesis (O'Rahilly & Müller, 2006), these findings are relevant to our hypothesis (this is not to say that savants may not suffer brain damage later in life, due to accident or disease). Second, an interesting finding is that many savants were born prematurely (Treffert, 2000). Although the exact reason why babies are born prematurely is often unknown, it is known that premature birth is usually related to disturbances during early organogenesis (Galis et al., 2006). So the combination of premature birth and savant syndrome suggest disturbances during early organogenesis. Third, it is important to establish that savant syndrome co-occurs with physical anomalies, besides brain anomalies, that are known to be established during early organogenesis, such as extra digits. Although a systematic study of physical anomalies in savants has yet to be conducted, from carefully reading of case histories, it emerges that many savants have physical anomalies. Treffert (2000) provides an overview of 90 savants. Some cases are described in great detail; others in a few sentences. Treffert mentioned physical abnormalities which we have listed in Table 3.1. Some savants have more than one physical abnormality. It should be noted that Treffert did not entertain any explicit hypothesis concerning the co-occurrence of savant syndrome and physical anomalies, so the anomalies listed in Table 3.1 are not the result of a systematic study. A systematic study of physical anomalies in savants would presumably result in a more extensive list.

Fourth, an important finding for our hypothesis is that savant syndrome co-occurs with Williams syndrome (Gillberg & Rasmussen, 1994), a genetic disorder characterized by deviant facial features and a heart defect. The origin of this disorder lies in abnormalities in neural crest development, which occurs during early organogenesis.

*Table 3.1 Physical anomalies in savants (n = 90), as mentioned in Treffert (2000)*

<b>Physical anomaly</b>	<b>Number of savants with anomaly</b>
Blindness or other visual defect	23
Motor disorder or delay	10
Disproportions in heads	7
Epilepsy	6
Deafness or other hearing defect	4
Disproportions in hands	3
Mute	3
High-arched feet	2
High-arched palate	2
Diabetes	1
Hypothyroid	1

Fifth, indirect evidence is provided by findings that characteristics that frequently co-occur with savant syndrome, such as autism and delayed language, have also been reported to co-occur with having an extra digit (Kumandas et al., 2004), other congenital abnormalities (Arndt et al., 2005; Miller et al., 2005; Wier et al., 2006) and minor malformations (Rodier et al., 1997) that are known to be established during early organogenesis. The co-occurrence of congenital abnormalities and characteristics of the savant syndrome provides support for the hypothesis that the savant syndrome is the result of pleiotropic effects in early embryological development. For example, autism co-occurs with Down syndrome or trisomy 21 (Ghaziuddin, 1997) and Patau syndrome or trisomy 13 (Konstantareas & Homatidis, 1999), i.e., two chromosomal abnormalities that have been reported to co-occur with an



increase in digit number (Castilla et al., 1998). This evidence is consistent with the hypothesis that the savant syndrome did not spread in the population due to pleiotropic effects in the early organogenesis stage.

## **Discussion**

We hypothesize that the failure of the savant syndrome to become established in the population through natural selection is due to a developmental constraint, as the concept is used in biology. The developmental constraint has its origins in the negative side-effects that are induced during the early organogenesis stage. These nullify the positive, potentially fitness enhancing, aspects of the savant syndrome. As we have only considered a few lines of support, further research is clearly necessary to test this hypothesis. This research should include a systematic assessment of physical anomalies that are known to be established in the early organogenesis stage in savants. These physical anomalies include, among others, polydactyly, midline disorders, such as cleft lip and spina bifida, congenital heart defects, the absence or incomplete development of one or more kidneys, or ear tags (Opitz, 1985). This research would also have clinical relevance, as the presence of physical anomalies might lead to more alertness towards severe psychiatric disorders, such as autism.

We have also shown that the biological conceptualization of a developmental constraint is relevant to psychiatric research. It is well known that if a mutated gene becomes active in the early organogenesis stage, its effects usually concern many phenotypic traits (Galis & Metz, 2001). In chapter 4 and 5, we examine the effects of mutations during the early organogenesis stage on the development of schizophrenia and autism. The vulnerability of the early organogenesis stage is the source of many disorders, both psychiatric and physical.

A remaining question is how disturbances in early embryological development may lead to psychiatric disorders that appear later in life. It is well-known that prenatal influences are correlated with the development of disorders such as autism (Glasson et al., 2004) and schizophrenia (M.C. Clarke et al., 2006). But can disturbances in embryological development explain the age of onset of disorders? Why is autism diagnosed as early as 18 months, while schizophrenia usually appears in early adulthood? Because autism and other developmental disorders are multigenic, this question is hard to answer. These disorders are

the result of multiple interactions among the effects of genes during both prenatal and postnatal life, leading to complex disorders with many characteristics. The problem of unraveling the influences of genes and interactions among genes on the development of disorders remains to be addressed. In the case of schizophrenia, there are already some findings that point in the direction of a specific gene, ZNF74, that is related to the age of onset of schizophrenia (Takase et al., 2001). For research on savants, it would be most interesting to get similar findings regarding autism.