Psychiatr. Pol. ONLINE FIRST Nr 317: 1–11
Published ahead of print 17 June 2023
www.psychiatriapolska.pl
ISSN 0033-2674 (PRINT), ISSN 2391-5854 (ONLINE)
DOI: https://doi.org/10.12740/PP/OnlineFirst/157104

An outline of savant syndrome

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Summary

This article is a review of research on savant syndrome from years 2000–2022 – available in PubMed, ResearchGate and Google Scholar databases.

Savant syndrome occurs with a frequency of 1.4 per 1,000 people with intellectual disabilities and in 10% of people with a diagnosis of autism spectrum disorders. Autism is the disorder that most often co-occurs with savant syndrome – in about 50% of all people with a savant syndrome diagnosis. Researchers distinguish between: congenital savant syndrome – which affects about 90% of people with the syndrome, and acquired savant syndrome – which occurs in the course of frontotemporal dementia or in people who have experienced trauma to the central nervous system. There are many cognitive theories that explain savant abilities. The anatomical basis of this entity has not been discovered using neuroimaging techniques.

To date, no unified theory has been created that reliably explains the etiology and pathomechanism of savant syndrome. Previous neuroimaging studies of people with symptoms of savant syndrome have been conducted on too few subjects. There are indications that magnetic brain stimulation may contribute to a better understanding of the neurophysiological basis of this syndrome.

Key words: savant syndrome, autism, neuroimaging

Introduction

Savant syndrome is a rare psychopathological syndrome. A person afflicted with this syndrome possesses extraordinary skills that are the opposite of his or her general limitations associated with the intellectual disability that is present in most cases. These skills, due to their nature, are called "islands of genius" [1]. The syndrome can be present from birth (congenital) or occur as a result of disease or injury to the central nervous system (acquired). Usually, the abilities relate to five general areas – art, calendar computation, music, mathematics, or visuospatial/mechanical skills. These

abilities usually occur individually [2]. 10% of people affected by autism spectrum disorders exhibit savant characteristics. People with symptoms of this syndrome experience many difficulties in communication, social skills. Savants are characterized by outstanding memory in narrow areas of special interest to them [3].

The first description of the syndrome was published in a German scientific journal in 1783 (*Gnōthi sautón oder Magazin zurr Erfahrungsseelenkunde als ein Lesebuch für Gelehrte und Ungelehrte*, Berlin) [4]. In 1887, Dr. J. Langdon Down described savant syndrome as a separate disease entity. He presented a description of a boy who could recite back and forth *The Fall of the Roman Empire*, verbatim [5].

Savants are characterized by a tendency to focus on details [6], possibly related to an obsessive personality profile [7]. It is worth mentioning that savant syndrome includes a spectrum of abilities of varying degrees. The rarest occurrence is characterized by the so-called prodigious savant. Treffner [8] estimated that in 2009 there were about 100 such prodigiously gifted savants in the world. The most common are "splinter skills". These are very specific skills disconnected from a related broader set of skills. They include obsessive memorization and interest in music trivia, sports, maps, and license plate numbers. "Talented savants" are people with cognitive disorders in whom musical, artistic or other special abilities are highly developed, usually in one area, and draw attention in juxtaposition with general disabilities [8].

Epidemiology

Treffert and Rebedew analyzed a group of 319 people with savant syndrome from 33 different countries (70% were from North America) [17]. They found that 90% of the group were savants with congenital abilities. Among congenital savants, the affliction was 4 times more common in men than in women. The most common skill of savants was musical aptitude. The occurrence of several skills simultaneously was present in 45% of cases, and a single skill was present in 55% of cases. Underlying disabilities in these individuals were autism spectrum disorders – in 75% of cases, while other central nervous system disorders were present in 25% of cases. Thus, not all people with savant syndrome also have autism spectrum disorders – about 50–75% of savants do not have autism, depending on the criteria adopted by researchers [2, 17].

Savant syndrome is estimated to occur in about one in a million people [41]. In turn, an assessment of the prevalence of savant syndrome in Finland showed that it occurs at a rate of 1.4 per 1,000 people with intellectual disabilities. It is worth mentioning that the most common ability was the ability to calculate calendar dates [9]. In 2004, Bölte and Poustka [16] reported that out of a study group of 254 people with idiopathic autism, 33 were savants, representing 13% of the group [16]. In 2009, Howlin et al. [27] found that in a group of 137 people with autism, 39 (28.5%) met criteria for savant syndrome, based on parental statements and cognitive tests. Of the

study group, one-third of the males exhibited some type of exceptional skills, compared to 19% of the females [27].

Autism is 4 times more common in males than in females, while savant syndrome is 6 times more common in males than in females [8]. An explanation for this fact can be sought in the work of Geschwind and Galaburd [28], who adopted a hypothesis that the right hemisphere completes development earlier than the left, and thus the left is more exposed prenatally to hormonal changes. High levels of testosterone in male fetuses can cause damage to the neurons of the left hemisphere of the brain, which can cause the right hemisphere to become dominant and thus develop exceptional skills [8]. There may also be other conditions resulting from impaired prenatal left hemisphere development, these include delayed speech, dyslexia and stuttering [8].

Classification attempts

It should be noted that not all savants have autistic disorders and not every autistic patient is a savant [2]. In general, about 10% of autistic people show savant abilities. About 50% of people with savant syndrome have autism, and the remaining 50% manifest other forms of developmental disorders or disabilities [42].

It is unclear why some autistic patients do not develop savant syndrome and others do. However, savant syndrome in adults with autism has been shown to have a unique cognitive profile in behavioral, cognitive and perceptual aspects that differs from adults with autism alone. This evidence was based on objective tests as well as self-report questionnaires. The distinguishing features of savant syndrome have not been found to be local information processing, social skills or increased pattern detection in calendar calculations [10].

Savants in some respects exhibit similar characteristics. They exhibited obsessive behavior, higher sensory sensitivity, greater systemization. and technical-spatial aptitude. In addition, savants showed a more engaged learning style and were more attentive when presented with a skill unfamiliar to them. These findings may in the future contribute to defining savant syndrome as a variant of autism [10]. Other studies report that children with autism and savant syndrome, compared to autistic individuals, show greater social reactivity. This has not been shown to be a family trait [11].

In addition to congenital savant syndrome, there is also an acquired variant. Treffert [12] described it as a situation in which, after damage or disease to the central nervous system, savant skills suddenly appear, sometimes at an astonishing level of sophistication, with such a skill not previously present in the person [12]. Treffert described several such cases, including the story of a 3-year-old boy who underwent meningitis. He then developed a remarkable musical talent. Another case tells of an 8-year-old boy who, after undergoing hemispherectomy surgery, acquired the ability to make calendar calculations extremely quickly. A 10-year-old boy developed a similar talent after being hit in the head by a baseball. In addition, Treffert [13] described the

case of two painters who experienced qualitative improvement in their painting skills after a stroke of the left occipital lobe and thalamus.

There are reports of patients with frontotemporal dementia who have acquired exceptional skills. Three patients have been described who became talented artists after developing frontotemporal dementia. Yet another similar case was described as developing "paradoxical functional facilitation," where "the loss of certain skills enables the emergence of others," such as artistic skills [13, p. 336–337]. Treffert [14] mentioned a researcher who collected 12 cases of frontotemporal dementia following which, patients developed artistic abilities. He also described an interesting case of a 51-year-old patient who suffered a subarachnoid hemorrhage associated with a cerebral artery aneurysm, and after the vascular incident, exceptional artistic talents were revealed.

Extraordinary abilities of savants

It is interesting to note that the savant's skills generally fall into one of five categories:

1. Music – most often playing the piano, playing multiple instruments (even 22), as well as composing alone, without playing an instrument;
2. Mathematics – involving instant counting or the ability to calculate prime numbers while lacking other basic math skills;
3. Spatial or mechanical skills – including the ability to measure distances accurately without tools, the ability to create complex models or plans with meticulous accuracy, or mastery of map drafting;
4. Art – mainly drawing, creating sculptures or painting;
5. Calculation of the calendar – interestingly enough, for most people this is an unknown skill [8].

Memorization is a common feature of people with savant syndrome. It can be linked to a specific ability or be a separate trait. There are known cases of savants who memorized phone books, population statistics or bus schedules [15].

Other skills were reported less frequently: the ability to judge the passage of time without using a watch, knowledge of multiple languages, unusual sensory abilities in touch, smell or sight [8].

Among the 33 autistic savants, the most common abilities were outstanding memory (16), musical aptitude (6), arithmetic (5), reading (4), and visual-spatial (1) and drawing abilities (1). Except in 4 cases, all subjects had more than 1 unusual skill typical of savants [16]. In another study, in a group of 45 savants, the most common ability was calendar counting (28), followed by excellent memory (13), artistic ability (6), and only 3 savants showed musical aptitude. Of this group of savants, only 7 had more than one unique talent [9]. And in yet another study, from an analysis on a larger group

of 319 savants, it was found that musical aptitude was most prevalent (25%), followed by outstanding memory (20%) and artistic aptitude (19%). The savant's single ability was present in 55% of cases [17].

Mechanisms of savant syndrome development

Takahata and Kato [18] classified possible cognitive models of savant syndrome into 3 categories:

1. The paradoxical functional facilitation model, which offers a potential explanation for how pathological conditions in the brain lead to the development of extraordinary abilities. It emphasizes the role of reciprocal inhibitory interactions between cortical areas, particularly the prefrontal cortex and posterior brain areas [19].
2. The hypermnesia model, which assumes that savants' abilities stem from latent or pre-existing cognitive functions, such as memory. However, recent neuropsychological research suggests that savants cope with problems using a strategy that differs significantly from that of non-autistic individuals [18]. This model is supported by the work of Miler [20], Pring and Hermelin [21]. They argued that there may be no differences between normal individuals and savants, in terms of the mental structure underlying exceptional abilities. It is also unclear whether savants have exceptional motivational factors or distinctive cognitive strengths. So, the skills manifested by savants share many characteristics with those of non-savants [20].
3. The autistic model mentions insufficient connectivity or interference with long-range fiber connections and emphasizes poor central coherence. Instead it associates local regions with increased intact connections with better local connectivity [18].

Some authors have argued that obsessive preoccupation may result from poor central coherence [22], while inefficiencies in executive control mechanisms may cause overuse of cognitive processing [7]. The authors proposed that the increased local connectivity is due to specialized and facilitated cognitive processes responsible for savant abilities [13]. There is a hypothesis suggesting that savants remain closed to one stimulus. It explains the deficiencies in savants in shifting selective attention from one stimulus to another one [23].

There are works that say savants have ready access to a lexicon of images due to better pictorial memory [24]. Goldberg's theory [25] is that savants' declarative memory is intact, even though their procedural memory remains dysfunctional. Another theory, the theory of enhanced perceptual functioning, assumes the presence of locally oriented auditory and visual perception and greater use of the posterior network in complex visual tasks. It includes the autonomy of information processing toward higher-order operations from low-level [26].

The pathophysiological basis of this phenomenon

There are scientific reports according to which, savant syndrome is associated with changes in brain metabolism resulting in altered brain network connections. These changes lead to inhibition of the left hemisphere of the brain, so that the right hemisphere can develop savant skills. Using neuromodulation techniques such as transcranial direct current stimulation and transcranial magnetic stimulation, changes of a similar type can be achieved. With the changes induced with their help, it may soon be possible to discover hidden human talents [29].

In 1998, Miller et al. [30] described five patients suffering from frontotemporal dementia who developed artistic abilities early in the disease. The patients underwent SPECT (Single Photon Emission Tomography). 4 patients had the temporal variant of frontotemporal dementia, in which the anterior temporal lobe was involved. In contrast, the dorsolateral frontal cortex was unaffected. Social and language skills were significantly impaired, but visual skills were preserved. These patients became artists whose work showed many common features but was original in its own way. Their creativity and output were based on visuality rather than verbality. Photographs, paintings were copies of reality devoid of a symbolic or abstract component. Miller et al. [30] put forward the thesis that loss of function of the frontotemporal lobes could lead to "facilitation" of the development of artistic abilities.

It seems that stimulation of the left frontotemporal area of the brain with repetitive transcranial magnetic pulses, makes it possible to block left hemisphere dominance [13] and enables the right hemisphere to develop skills similar to those found in savant syndrome [13]. In a study conducted by Young et al. [31], it was found that in 5 of the 17 participants in the study, the development of skills typical of the savant, i.e., declarative memory, drawing and calendar calculations, was noted during periodic stimulation. However, effective stimulation of the frontotemporal region can lead to deterioration of short-term memory function [31]. In 2003, Snyder et al. [32] conducted a similar study in which 11 participants underwent stimulation with guided low-frequency magnetic pulses to the left frontotemporal lobe. Significant improvements in drawing skills were observed in 4 patients. A chunk of the participants also showed improved proofreading skills [32]. Three years later, Snyder et al. [33] conducted another study in which they found that ten participants improved their ability to accurately guess the number of discrete objects, which occurred immediately after stimulation.

One study attempted to elucidate a potential cellular mechanism underlying savant syndrome. In a 13-year-old savant with a remarkable memory and autism spectrum disorder, a group of researchers generated induced pluripotent stem cells derived from urine (UiPSCs). The UiPSC-derived neurons showed increased expression levels of autism spectrum disorder (ASD) genes associated with learning disabilities, namely *TBR1*, *PAX6* and *FOXP2*. In addition, this was accompanied by an increased frequency

of spontaneous excitatory postsynaptic currents, increased size and mixed density of dendritic spines, and hypertrophic nerve cell bodies [34].

According to Snyder [35], savants have access to less processed information before it is given labels of meaning and before it is framed into holistic concepts. Thanks to top-down inhibition disorder, they can access information that is in all parts of the brain, but usually remains outside of consciousness. Hence, autistic savants are uncharacteristically literal with a tendency to focus more on the parts than the whole, which is beneficial in solving specific problems that require breaking cognitive patterns.

The place of anatomy and diagnostic imaging

Corrigan et al. [36] performed neuroimaging of the brain of a 63-year-old artistically gifted savant in 2012. No significant anatomical differences were observed to sufficiently explain the morphological basis of the unusual abilities. According to the morphological assessment, the brain volume (1,362 ml) was larger than the average value for an adult male. The right hemisphere of the brain was 1.9% larger. Compared to the left side, the right amygdala and right caudate nuclei were 24% and 9.9% larger. In contrast, the putamen on the left side was 8.3% larger. Both the left and right hippocampus had significantly increased axial and mean diffusivity compared to a comparison sample of adult males. The data revealed from this case, compared to a control sample, may underscore differences in the integrity of brain circuits involving these structures. In the parietal lobe, magnetic resonance spectroscopy showed markedly reduced concentrations of γ-aminobutyric acid and glutamate. Fractional anisotropy, on the other hand, showed a larger volume of nerve fibers on the right side. The brain assessment presented in this study, can be used to evaluate a general group of savants, to understand the source of unusual abilities.

Using functional magnetic resonance imaging, an effort was made to clarify whether savants with the ability to calculate the calendar (the ability to determine the day of the week for any date found in the calendar of any year) use calculations to answer the date question. Two savants were subjected to a specific test. They found increased activation of the parietal area for both date and arithmetic queries, with the area showing greater activity for more distant dates. This may be related to a characteristic of many calendar savants, who show increased response times for more distant dates. These results suggest that the calendar skills observed in savants are the result of intensive computational practice used in solving arithmetic questions [37]. The question remains as to how and why savants perform these activities.

Another study confirmed that the left frontal cortex, left middle temporal lobe and left hippocampus were involved in calendar calculations. These results suggest that extraordinary ability may be supported by memory processing. Since the brain circuitry involved in this unusual ability is very similar to that normally used for memory functions [38].

Loui et al. [39] conducted the study using tractography (a magnetic resonance imaging technique that allows in vivo visualization of the continuity and direction of nerve fibers) and the diffusion tensor imaging method (the use of specific MRI sequences to map the diffusion process of molecules, mainly water, in biological tissues). In this way, they examined the central nervous system of a musician endowed with absolute pitch. They observed that there was an increased number of nerve connections in his two temporal lobes, specifically in the upper parts. This is probably related to the presence of absolute pitch in this musician. In addition, the increased volume of pathways connecting the left middle temporal lobe to the left superior temporal lobe predicted the possession of absolute pitch. These findings may facilitate an understanding of the basis of savant syndrome associated with increased neuronal connectivity [39]. This hyperconnectivity may be related to synesthesia (the ability to associate sensations received by different senses with each other), which is often found in autism and savant syndrome. Six genes have been discovered that are associated with axon formation and are expressed in early childhood, when synesthesia develops. These genes are *COL4A1*, *ITGA2*, *MYO10*, *ROBO3*, *SLC9A6*, and *SLIT2* [40].

Recapitulation

Although savant syndrome was described in the medical literature more than 200 years ago, it is still not fully understood. The available data in the medical literature, are incomplete and in many aspects also contradictory. The most consistent reports have been found on the level of epidemiology describing the syndrome. The biggest obstacle to understanding the phenomenon seems to be the small number of people studied. Perhaps subjecting a larger number of subjects to modern neuroimaging techniques, such as tractography or magnetic resonance imaging, will make it possible to find commonalities, which will contribute to a better understanding of the syndrome. However, the greatest progress could be made by creating a unified and international savant research program. This would ensure that research results could be compared and that more people could be included in the study.

Perhaps in the future, more questions about this unusual syndrome will be answered. Through the use of modern research, we will learn the source behind these extraordinary abilities.

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Otrzymano: 11.08.2022
Zrecenzowano: 15.09.2022
Otrzymano po poprawie: 13.11.2022
Przyjęto do druku: 3.12.2022