Some Rare Neuropsychological Syndromes: Central Achromatopsia, Bálint’s Syndrome, Pure Word-Deafness, Supplementary Motor Area Aphasia
Alfredo Ardila
Online First Publication, June 29, 2017. http://dx.doi.org/10.1037/pne0000093

CITATION
Some Rare Neuropsychological Syndromes: Central Achromatopsia, Bálint’s Syndrome, Pure Word-Deafness, Supplementary Motor Area Aphasia

Alfredo Ardila
Florida International University

There are some unusual neuropsychological syndromes rarely reported in neuropsychological literature. This article presents a review of four of these unusual clinical syndromes: (a) Central achromatopsia (loss in the perception of colors due to a lesion of the visual association cortex); (b) Bálint’s syndrome (simultanagnosia, optic ataxia and gaze apraxia associated with bilateral parietal-occipital pathology); (c) pure word-deafness (clinical syndrome characterized by an inability to understand spoken language with preserved speech production and reading ability); and (d) aphasias of the supplementary motor area (significant difficulty in initiating and maintaining speech, regardless if the patient makes significant effort to speak due to a damage in the left supplementary motor area). It is concluded that although these syndromes are rare, it is fundamental to consider them to understand the brain organization of cognition. Their understanding is also crucial in the clinical analysis of patients with brain pathologies.

Keywords: central achromatopsia, pure word-deafness, SMA aphasia, Bálint’s syndrome

There exist unusual neuropsychological syndromes that are rarely reported in the neuropsychological literature. A previous article analyzed somatoparaphrenia, akinetopsia, reduplicative paramnesias, and autotopagnosia as examples of these unusual clinical syndromes (Ardila, 2016). In this article, central achromatopsia, pure word-deafness, aphasia of the supplementary motor area (SMA), and Bálint’s syndrome will be reviewed. They are so rare, that they are not even mentioned in basic neuropsychology textbooks (e.g., Horton & Wadding, 2008); or barely one or two of them are briefly mentioned (e.g., Banich, 2004; Baumont, 2008; Elias & Saucier, 2006; Heilman & Valenstein, 2012; Kolb & Whishaw, 2015; Rains, 2001; Zillmer & Spiers, 2001). Indeed, only some of these syndromes appear in neuropsychology dictionaries (Beaumont, Kenealy, & Rogers, 1999; Goodwin, 1989; Loring & Meador, 1999). Because of their rarity, from the fundamental point of view, they have a special interest for understanding the brain organization of cognition. From the clinical perspective, they may be overlooked if we are not sufficiently aware of their existence.

In this article, two visual perceptual syndromes (central achromatopsia and Bálint’s syndrome) and two language syndromes (pure word-deafness and aphasia of the SMA) are analyzed.

Central Achromatopsia

Definition and Initial Description

Central (or cerebral) achromatopsia has been defined as a loss in the perception of colors due to a lesion of the visual association cortex (Damasio, Yamada, Damasio, Corbett, & McKee, 1980; Meadows, 1974). Total loss of color perception due to a brain pathology is a very unusual clinical syndrome, and as a matter of fact, only few cases have reported this (Ardila, 2009; Bartolomeo, Bouchard-Lévi, & de Schotten, 2014; Cowey, Heywood, & Irving-Bell, 2001; Crognale, Duncan, Shoenard, Peterson, & Berryhill, 2013; Damasio et al., 1980; Fine & Parker, 1996; Jaeger, Krastel, & Braun, 1989; Kraft et al., 2014; Meadows, 1974; Mullin et al., 2001).
Some studies have shown the patient with central achromatopsia is capable of keeping the memory (imagination) of the colors, regardless the inability to perceive them (Shuren et al., 1996). As a matter of fact, the inability to get a mental representation of colors (meaning, knowing what colors are associated with which objects) corresponds more so to a color agnosia and not to an achromatopsia (De Vreese, 1991).

Central achromatopsia is usually correlated with other syndromes, such as prosopagnosia (Ardila, 2009; Damasio et al., 1980; Vaina, 1994), agnosia and topographical amnesia (Ardila, 2009; Kennard et al., 1995), visual agnosia (Ardila, 2009; Rizzo et al., 1993). In other words, defects in depth perception and spatial localization of stimuli (Rizzo et al., 1993), and pure alexia (Damasio, & Damasio, 1986; Rizzo et al., 1993; Shuren et al., 1996).

Ardila (2009) reported the case of a 47-year-old man who presented a cerebral achromatopsia associated with bilateral temporo-occipital infarcts, with a conservation of the imagination of colors (e.g., the patients easily and correctly answered the question, “What colors have the national flag?”). Additionally, the patient presented visual agnosia, he reported that “all the things looked alike”; for instance, he confuses a package of cigarettes with the TV control and was unable to recognize the streets around his house. When meeting a neighbor, he had difficulties recognizing her and could only identify her by her voice timbre. No defects in cranial nerves were found, but the patient reported he could only see tones of gray; no visual field defects were recorded; no motor or sensory defects were observed. Furthermore, the visual track was normal and no extinction with double visual stimulation was found. One week later, a significant recovery of achromatopsia and associated visual agnosia was found.

How This Syndrome Provides Further Insight Into the Brain

Evidently, this syndrome is crucial to understand the brain organization of visual perception, particularly color perception.

Conclusion

Disturbances in color recognition or achromatopsia can be due to both retina abnormalities, such as observed in Daltonism, and cortical pathology on central or cortical achromatopsia.

Bálint’s Syndrome

Definition and Initial Description

Bálint (1909) described a cortical syndrome characterized by, (a) “psychic paralysis of gaze,” in other words, difficulties in simultaneously processing multiple items (simultanagnosia); (b) “optic ataxia,” referring to deficits in visually guided limb and eye movements; and (c) “spatial disturbances of attention,” meaning defects in visual attention (gaze apraxia). As secondary to the attention impairments, the patient also presented defects in estimating distances; according to De Renzi (1982), these defects would be considered the fourth characteristic of this syndrome. Holmes (1918) described six similar cases, but included the frozen gaze phenomenon. This syndrome has been known as “Bálint’s syndrome” or “Bálint–Holmes’s syndrome.”

Simultanagnosia refers to the inability to perceive more than one single object at the time.
The term *simultanagnosia* was initially proposed by first Wolpert (1924) to describe a condition where the patient could see individual details of a complex scene, but could not grasp the overall meaning of the image. *Optic ataxia* (misreaching to visual targets) refers to impairment in visually guided reaching movements; it is a defect in guiding a finger toward a target, in absence of motor or sensory defects. It is consequently an impairment in the direction and visual control of movements. *Spatial disturbances of attention* (*Gaze apraxia*) refers to the patient’s inability to carry out voluntary eye movements and to keep the eyes fixed in a stimulus. Eye movements are somewhat random, and the patient has difficulties to look at a new stimulus.

Bálint’s syndrome is frequently associated with other disturbances, such as alexia without agraphia, visual agnosia, prosopagnosia, and visuospatial working memory defects (Ardila & Rosselli, 2007; Funayama, Nakagawa, & Sunagawa, 2015). Noteworthy, patients with Bálint’s syndrome frequently behave as if they were blind.

**Anatomical Correlates**

Since the initial descriptions presented by Bálint and Holmes, it has been accepted that Bálint’s syndrome is observed in cases of bilateral lesions; in the majority of the cases, parietal-occipital pathology (e.g., Rousseaux, Delafosse, Devos, Quint, & Lesoin, 1986; Valenza, Murray, Ptak, & Vuilleumier, 2004). Seemingly, to have a complete Bálint’s syndrome with the three clinical signs, bilateral lesions are required (De Renzi, 1982). A partial or incomplete Bálint’s syndrome can be observed with unilateral (usually left) pathology (Hécaen & Albert, 1978). This has also been observed in some specific pathologies such as Alzheimer’s disease (Mendez, Turner, Gilmore, Remler, & Tomsak, 1990) and Creutzfeldt–Jakob disease (Ances et al., 2004).

**Some Additional Studies**

Bálint’s syndrome has been considered as a particularly interesting clinical syndrome and different review articles of Bálint’s syndrome are currently available (e.g., Biotti, Pisella, & Vighetto, 2012; Chechlacz, & Humphreys, 2014; Mevorach et al., 2014).

In the neuropsychology literature, there is at least one case of rehabilitation of Bálint’s syndrome (Rosselli, Ardila, & Beltran, 2001). The rehabilitation protocol included both visuo-perceptual retraining and a functional program designed to provide rehabilitation in contexts that are meaningful to the patient. After 1 year of treatment, a second neuropsychological evaluation was carried out. It was found a significant improvement in terms of both objective testing and the return of an integrated and productive lifestyle.

**How This Syndrome Provides Further Insight Into the Brain**

Bálint’s syndrome represents one of the clinical syndromes which has most contributed to the understanding of visual and spatial perception (De Renzi, 1982; Farah, 2004; Hécaen & Albert, 1978).

**Conclusion**

Regardless that Bálint’s syndrome was reported over a century ago, the number of reported cases remains low; but its significance in understanding visual/spatial perception and integration has been particularly important.

**Pure Word-Deafness**

Pure word-deafness is an unusual clinical syndrome first described by Kussmaul in 1877, where a patient was unable to understand spoken language with preserved speech production and reading ability. He referred to this syndrome as “reinen Wortaubheit” (which translates as “pure word-deafness”). This syndrome is not equivalent to “word-deafness,” characterized by the impairments in phoneme discrimination, which can appear together with other language defects in Wernicke’s aphasia (Auerbach, Allard, Naeser, Alexander, & Albert, 1982). Sometimes is has been named as “auditory verbal agnosia” (Ulrich, 1978), but most authors prefer it as the original term “pure word-deafness” (e.g., Ackermann & Mathiak, 1999; Denes & Semenza, 1975; Gutschalk et al., 2015; Jörgens et al., 2008; Poeppe, 2001; Takahashi et al., 1992; Wolberg, Temlett, & Fritz, 1990).
There are some authors, however, who have proposed that “pure word-deafness” and “word-deafness” can be interpreted as variations of the same syndrome. Auerbach et al. (1982) suggested that there are two distinct subtypes of pure word deafness: (a) when the deficit is prephonemic and related to a temporal auditory acuity disorder and (b) as a form that is independent of a temporal auditory acuity disorder, and has a deficit in linguistic discrimination that does not adhere to a prephonemic pattern. The former would be associated with bilateral temporal lobe lesions, whereas the latter would be observed in cases of left unilateral lesions. In addition, the first form represents an apperceptive defect, whereas the second is a disorder in phonemic discrimination and may be considered a fragment of Wernicke’s aphasia. This distinction, however, has not really been explicitly integrated into aphasia literature. Reviewing the published cases, nonetheless, effectively makes it seem that, in some cases, an auditory acuity prephonemic disorder could exist (e.g., in those cases due to mesencephalic lesions; Kimiskidis et al., 2004; Vitte et al., 2002). Whereas in other cases, the defect seems to be a more exact phoneme discrimination impairment, as it was assumed in the original description of the syndrome (Kussmaul, 1877). Moreover, this condition is usually reported in cases of cortical temporal lesions (e.g., Rosati et al., 1982).

Most of the published cases of pure word-deafness are single case reports (e.g., Baddeley & Wilson, 1993; Gibbons, Oken, & Fried-Oken, 2012; Gutschalk et al., 2015; Jörgens et al., 2008; Kim et al., 2011; Notoya, Suzuki, Furukawa, & Enokido, 1991; Otsuki, Soma, Sato, Homma, & Tsuji, 1998; Praamstra, Ha-goort, Maassen, & Crul, 1991; Slevc, Martin, Hamilton, & Joannis, 2011; Takahashi et al., 1992; Tanaka, Yamadori & Mori, 1987), and only occasionally the report of two cases (e.g., Hayashi & Hayashi, 2007; Shivashankar et al., 2001; Vitte et al., 2002). This paucity in reports is due to its rarity.

According to the original description presented by Kussmaul (1877), the core distinguishing characteristics are presented as a severe inability to understand spoken language with preserved speech production, discrimination of natural sounds, and reading ability. Patients with this syndrome have extreme difficulties discriminating phonemic contrasts. As a consequence, they exhibit a profound speech perception deficit with subsequent auditory language comprehension impairment. Speech can be perceived as a whisper; or as if other people were speaking in a foreign unknown language. In many features, this problem compares with the altered speech perception of late bilinguals, unable to discriminate non-native phonemes (Feldmann, 2004). However, the patient can read and understand written language, demonstrating that the language disturbance is limited to the auditory perception of language phonemes. It must also be emphasized that speech production is preserved, and hence, the lexical, semantic and grammatical knowledge of the language are not impaired; it is only the phoneme auditory perception.

Furthermore, the majority of word-deafness reported cases are due to lesions of the superior left temporal lobe including primary auditory area (Brodmann Areas 41 and 42; e.g., Auerbach et al., 1982; Gutschalk et al., 2015; Stefanatos, Gershkoff, and Madigan (2005)), suggesting a purely perceptual deficit, and hence, an auditory (verbal) agnosia. However, some additional localizations of the pathology associated with this syndrome have been reported; Joswig, Schönenberger, Brügge, Richter, and Surbeck (2015) reported a case of pure word deafness, associated with a pineal germinoma on the inferior colliculi in a young patient. After percutaneous radiation therapy, the size of the tumor decreased significantly, but the audiometry demonstrated a complete regression of the auditory deficit. In addition, there are at least three additional cases of the syndrome caused by a mesencephalic lesion (Kimiskidis et al., 2004; Vitte et al., 2002). Hayashi and Hayashi (2007) studied two patients with pure word-deafness due to a subcortical hemorrhage in the left temporal lobe. There are also reports of pure word-deafness associated with bilateral(Earnest, Monroe, & Yarnell, 1977; Tanaka et al., 1987) or left unilateral temporoparietal lesions (Sato, Yasui, Isobe, & Kobayashi, 1982; Slevc et al., 2011; Takahashi et al., 1992).

Pure word-deafness has also been reported in some particular clinical syndromes such as dementia, both in Alzheimer’s disease (Kim et al., 2011), and in frontotemporal dementia (Gibbons et al., 2012)– and Landau-Kleffner syndrome (Notoya et al., 1991; Stefanatos, 2008).
Pure word-deafness has been reported as a subtype of primary progressive aphasia (Otsuki et al., 1998; Jörgens et al., 2008).

**Aphasia of the SMA**

**Definition and Initial Description**

Brickner (1940) reported that electrocortical stimulation of SMA resulted in continuous perseveration. Penfield and Welch (1951) observed arrest of speech associated with stimulation of this cortical region. However, language disturbances associated with SMA pathology were reported relatively late in the aphasia literature. Arseni and Botez (1961) reported speech disturbances caused by tumors of the supplementary motor area. The specific clinical characteristics of this type of aphasia were later described by Rubens (1975, 1976); he studied two right-handed patients with evidence of major infarction in the territory of the left anterior cerebral artery; according to Rubens, these patients presented: (a) a significant dissociation between intact repetition and grossly disturbed spontaneous conversational speech, (b) an absence of phonemic paraphasias, (c) a lack of speech inhibition, and (d) relative preservation of confrontation naming and comprehension. The author points out that, despite the initially profound motor aphasia, spontaneous conversational speech returned in two to three months. Later on, Jonas (1981) referred to the participation of the SMA in speech emission.

**Anatomical Correlates**

The occlusion of the left anterior cerebral artery is the most frequent etiology of this aphasia, but has also been reported in cases of tumors and traumatic head injury (e.g., Ardila & Lopez, 1984; Krainik et al., 2003). Speech is characterized by (a) an initial mutism lasting about 2–10 days; (b) later, a virtually total inability to initiate speech; (c) nearly normal speech repetition; (d) a normal language understanding; and (e) absence of echolalia. A right leg paresis and right leg sensory loss are observed; a mild right shoulder paresis and Babinski sign are also found. Language recovery is outstanding and is usually observed during the following few weeks or months.

The SMA is a mesial premotor area involved in the ability to sequence multiple movements performed in a particular order (Tanji & Shima, 1994). SMA participates in initiating, maintaining, coordinating, and planning complex sequences of movements (Alario, Chainay, Lehericy, & Cohen, 2006); it receives information from the posterior parietal and frontal association areas, and projects it to the primary motor cortex (Kandel, Schwartz, & Jessell, 1995). SMA damage is also associated with slow reaction time (RT; Alexander, Stuss, Picton, Shallice, & Gillingham, 2007). It has been observed that activation of the SMA precedes voluntary movement (Erdler et al., 2000); therefore, it can be assumed that SMA plays crucial role in the motor expression of speech processing (Fried et al., 1991). Nonetheless, the SMA is located at some distance—and indeed far away—from the classic language area postulated by Dejerine (1914) and assumed in most anatomical models of aphasia.

It has been suggested that SMA has a close connectional relationship with the prefrontal cortex and plays a critical role in the update of verbal representations (Tanaka, Honda, & Sado, 2005). Neuroimaging studies in humans have demonstrated that SMA is active not only when speaking, but also when performing diverse cognitive tasks, such as spatial working memory (Jonides et al., 1993), verbal working memory (Paulesu, Frith, & Frackowiak, 1993), arithmetic tasks (Hanakawa et al., 2002), spatial mental imagery (Mellet et al., 1996), and spatial attention (Simon et al., 2002) activities.

Evidently, the SMA is a complex motor cortical area, not primarily a language related brain area. Its role in language seemingly refers to the motor ability to initiate and maintain voluntary speech production.

**Some Additional Studies**

In the neurological and neuropsychological literature there exist only few publications about this type of aphasia. Most often, they are single case reports (e.g., Ardila & Lopez, 1984; Masdeu, Schoene, & Funkenstein, 1978; Pai, 1999; Ziegler, Kilian, & Deger, 1997). However, Freedman, Alexander, and Naeser (1984) described 15 patients with pathologies in the anterior cerebral artery of the dominant hemisphere involving the SMA—even though they used the name “transcortical motor aphasia”—. They described the clinical characteristics as limited spon-
taneous speech, intact repetition, normal articulation, and good auditory comprehension. Additionally, Krainik et al. (2003) studied 12 patients with surgical resection of medial frontal lesions, and attempted to relate the occurrence of speech deficits to the specific surgical lesion of the SMA. Six of 12 patients presented speech disorders. The deficit was similar in all patients, consisting of a global reduction in spontaneous speech, ranging from a complete mutism to a less severe speech reduction, which recovered within a few weeks or months.

The following language characteristics are usually found (Ardila, 2014): spontaneous language is limited, but language understanding and language repetition are normal; there is a significant difficulty in initiating and maintaining speech, regardless of the patient’s significant effort to speak; reading aloud is defective but reading understanding is nearly normal; writing is slow and painstaking.

Legs have a motor representation in the medial aspect of the frontal lobe motor areas. Consequently, right leg hemiparesis represents the most important neurological abnormality; frequently, an extension of the pathology found toward the parietal lobe, and hence, some right leg sensory loss can also be observed.

How This Syndrome Provides Further Insight into the Brain

Aphasia of the SMA is rarely reported in neurological and neuropsychological literature. As a type of aphasia, SMA presents some special characteristics, (a) appears as a consequence of a lesion outside the classical language area (i.e., perisylvian area of the left hemisphere); (b) was reported late in the history of aphasiology; and indeed was systematically described only during the mid1970s; and (c) tends to present a rapid spontaneous improvement; this last characteristic may be the reason to account why it was overlooked for so long time. Noteworthy, when initially described, SMA was interpreted as a transcortical motor aphasia (Rubens, 1976), and that idea remained for some time (e.g., Alexander & Schmitt, 1980; Freedman et al., 1984), regardless of the significant differences in location and clinical characteristics with the traditional transcortical motor aphasia (Ardila & Lopez, 1984; Berthier, 1999; Goldstein, 1917). Currently, it has been suggested that the aphasia of the SMA is somewhat secondary or “peripheral” aphasia, impairing the mechanisms required to produce speech, but not the language itself (Ardila, 2010).

Aphasia of the SMA illustrates the complex overlap between language and motor processes, which has contributed positively to further our understanding about brain representation of linguistic processes.

Conclusion

Aphasia of the SMA represents an unusual clinical syndrome characterized by the disturbance of initiating, maintaining, coordinating, and planning complex sequences of movements used in speech. It could be regarded as an aphasic or as a motor syndrome.

General Conclusion

In neuropsychology—as in any clinical area—there are some unusual syndromes that are found very sporadically. But their rarity does not diminish their importance in the fundamental understanding about the brain organization of cognition, as well as in clinical analysis of patients with brain pathologies. The four clinical syndromes analyzed before illustrate this type of neuropsychological unusual syndromes.

In this article, two visual perceptual syndromes (central achromatopsia and Bálint’s syndrome) and two language syndromes (pure word-deafness and aphasia of the SMA) were analyzed. Regardless their scarcity, their contribution to the understanding of the brain organization of visual perception and language has been enormous. It does not seem easy to understand the cerebral organization of visual perception (including perceptual integration, spatial location of objects, and recognition of color features) without considering the two perceptual syndromes analyzed in this article. It does not seem easy to understand brain language organization with regard both to its perception, as well as to its motor production without considering the neurological bases of phoneme perception and motor speech control.

References

Pathomechanismen zentraler Hörstörungen (reine Worttaubheit, auditive Agnosie, Rindentaubheit) [Symptomatology, pathological anomalies and mechanisms of central hearing disorders (pure word deafness, auditory agnosia)]. Fortschritte Neurolog · Psychiatr, 67, 509–523.


RARE SYNDROMES


Vitte, E., Tankéré, F., Bernat, I., Zouaoui, A., Lamas, G., & Soudant, J. (2002). Midbrain deafness with


Received January 26, 2017
Accepted May 29, 2017