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Neurophysiological responses to music and vibroacoustic stimuli in Rett syndrome

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ABSTRACT

People with Rett syndrome (RTT) have severe communicative difficulties. They have as well an immature brainstem that implies dysfunction of the autonomic nervous system. Music plays an important role in their life, is often used as a motivating tool in a variety of situations and activities, and caregivers are often clear about people with RTTs favourites. The aim of this study was to investigate physiological and emotional responses related to six different musical stimuli in people with RTT. The study included 29 participants with RTT who were referred to the Swedish Rett Center for medical brainstem assessment during the period 2006–2007. 11 children with a typical developmental pattern were used as comparison. A repeated measures design was used, and physiological data were collected from a neurophysiological brainstem assessment. The continuous dependent variables measured were Cardiac Vagal Tone (CVT), Cardiac Sensitivity to Baroreflex (CSB), Mean Arterial Blood Pressure (MAP) and the Coefficient of Variation of Mean Arterial Blood Pressure (MAP-CV). These parameters were used to categorise brainstem responses as parasympathetic (calming) response, sympathetic (activating) response, arousal (alerting) response and unclear response. The results showed that all participants responded to the musical stimuli, but not always in the expected way. It was noticeable that both people with and without RTT responded with an arousal to all musical stimuli to begin with. Even though the initial expressions sometimes changed after some time due to poor control functions of their brainstem, the present results are consistent with the possibility that the RTT participants' normal responses to music are intact. These findings may explain why music is so important for individuals with RTT throughout life.

1. Introduction

Rett syndrome (RTT) is a neurodevelopmental non-progressive disorder which is identified all over the world. The diagnosis includes intellectual disability. An apparently normal early development was one initial criterion for RTT. Various scientists nowadays consider RTT to be a neurodevelopmental disorder manifesting very soon after birth, even among variants, including preserved speech variants (Einspieler, Kerr, & Preechtl, 2005; Marschik et al., 2013). The developmental

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slowing-down starts at around 25 weeks of infancy, although there is considerable variation between individuals. There is no continuing deterioration in serial clinical examinations, no progressive alteration in Magnetic Resonance Imaging, and no evidence of progressive deterioration in EEG (Armstrong & Kinney, 2001). The classical form of RTT has an incidence of 1:10,000 newborn girls (Kerr & Witt Engerström, 2001). The Rett disorder almost exclusively affects females, but a few males have received the diagnosis (Leonard et al., 2001). It is considered to be a non-hereditary genetic disorder (less than 1%) in females. In summary, RTT is still primarily a clinical diagnosis that is strongly supported by a mutation in the MECP2 gene.

People with RTT have severe communication impairments, which is one of the criteria connected with the syndrome (Hagberg & Witt Engerström, 1986; Neul et al., 2010). The communication difficulties in RTT include loss of words and a strongly limited ability to use expressive language (Didden et al., 2010; Marschik et al., 2012; Sigafoos et al., 2011). Receptive language is preserved in varying degrees (Bartolotta, Zipp, Simpkins, & Glazewski, 2011; Marschik et al., 2013; Wine, 2009). Purposeful use of the hands is difficult because of involuntary stereotyped hand movements and dyspraxia. This affects the ability of people with RTT to actively communicate using pictures, computers, sign language and speech synthesisers. Consequently they are extremely dependent on the ability of caregivers to observe and interpret their poor communicative signals – including emotional and facial expressions. However, some facial expressions might emanate from abnormal spontaneous brainstem activity (ASBA) because of the immature brainstem (Bergström-Isacsson, Julu, & Witt Engerström, 2007; Bergström-Isacsson, Lagerkvist, Holck, & Gold, 2013; Julu, 2001). Although Bergström-Isacsson et al. (2013) claim that it *is* possible to identify facial expressions based on detailed and systematic observations, the abnormal brainstem activity complicates interpretations of facial expressions and emotions in the RTT population.

1.1. The autonomic nervous system

People with RTT have an immature brainstem that implies dysfunction of the autonomic nervous system, and it stays immature throughout life (Julu et al., 2008; Kerr & Witt Engerström, 2001; Porges, Doussard-Roosevelt, Portales, & Greenspan, 1996). The autonomic nervous system is part of the motor output of emotions (Bergström-Isacsson et al., 2007; Guyenet et al., 1996). It is made up of the sympathetic and the parasympathetic nervous systems, both of which are influenced, but not fully controlled, by consciousness (Julu, 2001).

The sympathetic activity, which is continuously active, has a close relationship with changes in the Mean Arterial Blood Pressure (MAP, measured in mmHg) (Sun & Guyenet, 1986). The sympathetic part increases blood pressure and pulse to regulate sudden changes. Changes in mean arterial pressure can be used as a non-invasive index of the brainstem's sympathetic activity, and it is possible to monitor mean arterial pressure beat by beat, using non-invasive methods. The sympathetic activity could be explained as an activating of the autonomic nervous system.

The parasympathetic nervous system is activated by afferent signals from different parts of the body, creating rest and calm in situations when the body's reserves are being replenished. The parasympathetic part functions as a natural brake that prevents the sympathetic part from getting out of control (e.g. a physiological condition of stress, including uncontrolled variability in breathing, pulse and blood pressure). Cardiac Vagal Tone (CVT measured in LVS) represents the brainstem parasympathetic activity. It is possible to monitor CVT continuously in real time by non-invasive methods (Julu et al., 2001). The parasympathetic activity could be explained as a calming response.

1.2. The connection between arousal, affects and emotions

Arousal is the fundamental force for all bodily and mental activities, and the autonomic nervous system is, together with the arousal system, the pathway into our emotions (Pfaff, 2006). Humans are born with affects such as anger, happiness and sadness, and connected with our survival (Damasio, 2010; Ekman, 1992). The arousal systems are located in the brainstem and the signals go from the brainstem to cortical centres and from the cortex and emotional centres to the arousal systems. They constantly regulate one another but they can act alone, e.g. an internal startle and momentary arousal peak. Arousal is part of the defence responses which means that an increased brainstem parasympathetic activity is similar to what happens in drowsiness (Delamont, Julu, & Jamal, 1998). An arousal in itself is neither positive nor negative, it is simply a physiological response (Stern, 2010). The experience following an arousal in a person depends on the stimulus, its interpretation and past memory of the stimulus, if any. Repeated experiences of responses to stress or pleasure are important for learning as they condition a person's behaviour towards a stimulation that is pleasant, and to avoid other stimulations that give unpleasant emotions (Pfaff, 2006).

As emotions refer to the *experiences* of affects, this is a key part of the process of an individual's interaction with stimuli. Facial, vocal, or gestural behaviour serves as an *indicator* of both affect and emotion. Emotions are present in our bodily consciousness, in memories, in our perception, in our relations with others and also as part of our communicative competence. They construct a meaning, irrespective of disability or normal development. Emotions can create support in the search for how to behave in the world, and in a person's understanding of it, both on a bodily and a verbal level.

1.3. Music and vibroacoustic stimuli

For people with RTT, music plays an important role in life, and most of them have clear musical preferences (Elefant, 2002; Houtaling, 2003; Merker, Bergström-Isacsson, & Witt Engerström, 2001; Wesecky, 1986). Music is often used