

Stereotypies and Movement disorders in Rett syndrome

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Patients with Rett syndrome evolve from a hyperkinetic to a hypokinetic state, and a large series of abnormal movements may be observed along their lives such as stereotypies, tremor, chorea, myoclonus, ataxia, dystonia, and rigidity.

Stereotypies have been defined as involuntary, coordinated, patterned, repetitive, often rhythmical, and no purposeful movements. Stereotypies in Rett syndrome are a diagnostic hallmark present in all stages of the disease. It is now accepted that hand stereotypies coincide with or sometimes precede the loss of purposeful hand movements in very early development of RTT girls (Einspieler et al. 2005, Temudo et al. 2007).

Early in life, even before regression, Rett syndrome patients demonstrate abnormal posture, tone, and movements. Using meticulous recordings with special attention paid to the face, the hands, and body movements, several authors have demonstrated an abnormal quality of general movements (100%), tongue protrusion (62%), postural stiffness (58%), asymmetric eye opening and closing (56%), abnormal finger movements (52%), hand stereotypies (42%), bursts of abnormal facial expressions (42%), bizarre smile (32%), tremor (28%), and stereotyped body movements (15%). (Nomura and Segawa 1990, Nomura and Segawa 1992, Einspieler et al. 2005, Nomura 2005)

Hand stereotypies can be in the midline, with symmetrical movements of both hands (washing, clapping, tapping, wringing, hand mouthing), or with hands apart, more frequently each hand performing a different movement (hair pulling with one hand, the other tapping the trunk; hair pulling with one hand, with the other to the mouth; pill-rolling with one hand; twitching of two fingers; castanets, etc.). These almost continuous, repetitive, and compulsive automatisms disappear during sleep and may aggravate with anxiety.

In addition to hand stereotypies, Rett patients also present stereotypies with other topographies: cervical retropulsion (bending the neck backward), head rolling, lip protrusion, eye rolling, trunk rocking, intermittent leg elevation, and tapping of the floor, toe walking, and swaying movements of all the body with shifting weight from one leg to the other. Stereotypies can also be very complex at the beginning of the disease, some girls exhibiting like a "stereotyped dance" (Temudo et al. 2007, Temudo et al. 2008).

However, because of their heterogeneity, varying in location, frequency, and severity, systematic assessment of hand stereotypies in Rett syndrome is challenging. (Dy et al. 2017). Very rarely, some hand stereotypies resemble choreoathetoid movements (FitzGerald et al. 1990).

Dystonia is also a frequent movement disorder in Rett syndrome (60% of patients), more often crural (involves leg or thigh) or generalized, but also focal involving the upper or lower limbs. Usually, dystonia is asymmetric, the right side being more affected. Scoliosis, a common feature of RD, mainly at its later stages, is considered to be a consequence of this postural asymmetry. (FitzGerald et al. 1990, Hagberg and Romell 2002) (Temudo et al. 2008).

Rigid-Akinetic Syndrome

An inexpressive, “mask-like,” face may be an early clinical sign of Rett disorder, when accompanied by disproportionate eye communicating ability and normal eye blinking. Hypomimia (a reduction in the expressiveness of the face), bradykinesia, and rigidity become more severe and common in older girls. Remarkably, patients with severe motor presentations, who never acquire an independent gait, present rigidity very early in the disease evolution (less than 5 years). (FitzGerald et al. 1990, Temudo et al. 2008)

Ataxic-Rigid Gait: Rett's Gait

The majority of the patients acquire independent ambulation. With the progression of the disease, gait becomes more rigid, with poor arm balance, and some patients exhibit a particular wide-based rigid gait with abdominal muscle contraction and hyperextension of the legs. They can also present with freezing when initiating a movement. (FitzGerald et al. 1990, Temudo et al. 2008, Humphreys and Barrowman 2016)

Evolution of stereotypies in adolescents and women with Rett syndrome (Vignoli et al. 2009)

The pattern of manual stereotypies is maintained lifelong, even if, as soon as patients become adult, the movement disorder shows slower components and is interfered by tremor. In some patients, stereotypies tend to become simpler and less severe with increasing age because of patient's rigidity (Temudo et al. 2008). A small percentage of women keep their manual ability (i.e., holding an object, drinking, and eating by themselves) in adult age.

Looking at their distribution, stereotypies most frequently involve hand and mouth (FitzGerald et al. 1990, Vignoli et al. 2009). Regarding frequency of stereotypies, they are constant during daytime while they disappear during sleep. All patients present with motor stereotypies involving separated or joined hands: most frequent movements are mouthing (50%), pill rolling, and twisting two or three fingers (50%), bruxism (50%), oro-facio-lingual movements (40%) and less frequently leg involvement or trunk rocking. Many adult patients present a tremor disorder, which appears later in the evolution of the disease. The mean frequency of tremor is around 5 Hz, thus overlapping mean frequency of tremor encountered in Parkinson's disease and significantly lower compared with other types of tremor, i.e., essential tremor (FitzGerald et al. 1990, Vignoli et al. 2009)

Effect of hand splints on stereotypic hand behaviour of girls with Rett syndrome:

Only three studies report the effects of hand splints on hand stereotypies in Rett syndrome. They consist in cuffs encircling the palm that position the subjects' thumbs in abduction. After application of hand splints, some patients show a decrease in the amount of time spent in stereotypic hand behaviour. Although splints have showed a positive effect on hand movements in Rett's syndrome, they could however lead to other, undesirable, movements. Whether splints have a positive effect on the functional use of the hand should thus be investigated in more subjects. (Naganuma and Billingsley 1988, Tuten and Miedaner 1989, Bumin et al. 2002)

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