

Title – Understanding Russell-Silver syndrome and its potential psychosocial impact

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Abstract

We are increasingly living in a society that values appearance and anyone that differs from the 'norm' or what is culturally defined as 'attractive' may experience psychological distress based on others reactions and perceptions as well as their own (Harcourt and Rumsey, 2012). People living with appearance altering congenital conditions may not be receiving the psychological support they need to increase confidence, self-esteem and build resilience to develop and retain healthy romantic relationships in adolescence and into adulthood. In this article we present the potential impact that looking different could have on people with Russell-Silver syndrome, a rare genetic condition which affects height and facial/body symmetry; as well as outline research currently being conducted in Southampton with adolescents and adults with the condition.

What is Russell-Silver syndrome and how it is treated?

Russell-Silver syndrome (RSS), also known as Silver-Russell syndrome, is genetic but not usually inherited (Binder et al., 2011). It is characterised by slow growth in the womb, poor post-natal growth, short stature in adulthood, triangular facial appearance with a broad prominent forehead and body/facial asymmetry (Wakeling, 2011). The prevalence of RSS is not easily determined (Wakeling, 2011), however, the condition is thought to affect 1 in every 100,000 people (US National Library of Medicine, 2008); males and females are equally affected (Binder et al., 2011). Currently there are two known molecular abnormalities that cause RSS: A loss of DNA methylation at the H19 locus causes 60% of cases (this epigenetic mutation causes a reduction in a growth promoting factor called IGF2), maternal uniparental disomy of chromosome 7 (MatUPD7, the inheritance of both chromosome 7's from the mother with no contribution from the father) causes 5-10% of cases; the remaining patients have clinical features but an unknown molecular cause (Wakeling, 2011).

Currently the primary treatment for RSS is growth hormone for short stature and, whilst there is evidence that GH treatment increases height in other growth conditions (Dahlgren, 2011), it is less clear how effective this treatment is for RSS. GH cannot treat other appearance differences such as facial or body asymmetry and might make it more prominent. Other treatment options include procedures for overcrowded teeth or leg lengthening surgery.

How does RSS affect appearance?

RSS affects appearance in a variety of ways, notably in short stature and asymmetry. Children often show slow growth throughout childhood coupled to reduced growth at birth and typically no catch-up growth during puberty resulting in a height -4.2 SD below the mean in adulthood, with an average height of 140cm (4' 7") in women and 151cm (4' 11") for men (Binder et al., 2011). Average height for men in the general population is 5'8" and for women it is 5'4" (Royal College of Paediatrics and Child Health, 2015). Asymmetry can

affect the body, face and/or limbs and prevalence varies from about one third upwards (Price et al., 1999; Wakeling et al., 2010).

The psychosocial impact of being visibly different with RSS

Being shorter than average may impact significantly on a person's life. Research has found that children with SS, compared to peers with normal stature, can often feel less satisfied with their appearance (Laub, 2012). Having a negative body image may result in lower self-esteem and in turn affect psychological wellbeing, causing anxiety and depression (Vilhjalmsson et al., 2012). SS can predict negative body image and is often found to be the case in boys more so than girls; problems can begin when an individual's body image deviates from the cultural ideal, for example, being a taller man rather than shorter is seen as more desirable as taller men may receive more favourable attention and therefore have more opportunities romantically and socially (ibid).

However, being short does not necessarily lead to negative psychosocial outcomes. Recent research from Schanke and Thorsen (2015) identified coping strategies that people may use to deal with psychosocial challenges. Ten people between 45-65 years old were interviewed and findings highlighted the important role of stigma-handling and resilience throughout the lifespan. The challenges and coping strategies of people with SS are beginning to be understood but the complexities require further exploration in order to help support those with the condition.

A further societal pressure stems from the widely held perception that symmetry is linked to beauty. Patients with RSS often experience visible asymmetry, for example, one side of their body could be visibly smaller than the other and weight gain may accentuate this difference further. Evidence shows that a preference for symmetrical faces may be related to sexual selection and partner choice for both men and women (Rhodes, 2006, Thornhill and Gangestad, 1999, Griffey and Little, 2014, Little et al., 2008) and may be viewed as an indicator of having healthy genes to pass onto offspring (Thornhill and Gangestad, 1999). Findings are similar for sexually dimorphic faces (that is women with feminine features and men with masculine features) (Rhodes, 2006, Grammer and Thornhill, 1994, Griffey and Little, 2014, Little et al., 2008) and for symmetrical bodies (Ganstead and Simpson, 2000). Having romantic relationships is one of the most important developmental experiences in

adolescent as they can improve self-esteem, emotional support and lead to the ability to form healthy relationships in adulthood (Sorensen, 2007). However, evidence which explores the impact of asymmetry on psychosocial adjustment and intimacy for patients with RSS is lacking, and is under researched in patients with other appearance altering conditions (see Sharratt 2015).

Enhancing treatment and psychosocial support for people with RSS

The Wessex Imprinting Group, from the University of Southampton, is conducting a study with people who have RSS syndrome called STAARS UK (Study of Adults and Adolescents with Russell-Silver Syndrome). The Child Growth Foundation, a UK charity for children and adults with growth and endocrine issues, has pressed for more information on health outcomes for adults with RSS syndrome as little research currently exists. The STAARS study has three main aims: first, to ascertain what the long-term outcomes are for people with RSS as there is significant evidence to link increased risk of heart disease (Barker et al., 1989, Barker et al., 1993, Barker, 2005, Hales et al., 1991, Osmond et al., 1993) and diabetes (Barker, 2005, Hales et al., 1991, Barker et al., 1993) in adulthood with growth problems in the womb and early childhood. The second aim concerns growth hormone, the only treatment currently offered to children and adolescents with RSS syndrome. The study aims to recruit 100 people with RSS syndrome to undertake clinical investigations to understand more about health outcomes. The study's third aim involves exploring the lived experience of people with RSS through in-depth interviews with a sub-set of the total sample.

In conclusion

People with RSS are living with a visibly different appearance and research shows looking different from the 'norm' may result in psychological distress. The in-depth interview aspect of the STAARS study may give an insight into how people with RSS feel about their appearance and what impact looking different has on them and their relationships. Currently, children and adolescents are not routinely screened for psychological issues or offered psychological support for appearance related concerns in endocrine clinics with clinicians focus on treating growth. All HCPs working with patients who have an appearance altering condition should be aware of the possible psychosocial impact on the development of social, and especially, romantic relationships.

4 summarising key sentences for the end of the article

- Having an appearance that differs from the 'norm' can negatively impact on a person's wellbeing and some people are more resilient than others
- Forming romantic relationships is an important part of development during adolescents and those with RSS may find this harder
- HCPs working with adolescent with appearance altering conditions, like RSS and other growth disorders, could ask questions about how their appearance affects different aspects of their lives
- The STAARS study will report their findings in 2016/17 and if you are interested in the outcomes please visit:

www.southampton.ac.uk/geneticimprinting/informationpatients/staars.page

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