

# Puberty in Rett syndrome

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Parents often find puberty a difficult time for all their children. In fact, it may turn out to be an easier transition for the person with Rett syndrome than for brothers or sisters. However, there are a number of specific issues that you may need to give particular attention to at this time.

## Menarche

The main event for girls entering teenage years is menarche (or starting periods). In most girls with Rett syndrome, this happens without any difficulty, although you should not be concerned if your daughter is late in starting. She is particularly likely to start late and / or to have irregular periods if she is underweight, and this may be an indicator to check her growth and possibly increase calories. Keep a chart of her periods and take note if she seems to be at all distressed – like many women she may need some simple painkillers for period pain.

## Epilepsy

This is an area where you may see improvement in adolescence. Epilepsy tends to get better with increasing age in Rett syndrome, so if you have had a prolonged fit-free period it may be worth discussing careful withdrawal of medication with the person with Rett syndrome's GP or consultant.

## Feeding and growth

A large proportion of people with Rett syndrome have difficulty chewing and swallowing, despite a voracious appetite. Parents usually manage to find the best food consistency to ensure adequate weight gain through early childhood, although sometimes food supplements or other feeding techniques have to be used.

However, it is very important to keep a close eye on eating skills and weight through adolescence, since a number of people with Rett syndrome go off their food, have more difficulty eating and/or fall into a cycle of weight loss and ill health. Careful monitoring of weight will alert you to early warning signs, and allow proactive management, which may include dietary supplements, changes in texture or delivery, and in a number of cases feeding by gastrostomy.

## Mobility/Scoliosis

Through adolescence, the person with Rett syndrome may begin to stiffen up. It is very important to try and maintain physiotherapy, hydrotherapy, horse riding and all other physical activities to minimise the chances of the person with Rett syndrome going off their feet or developing contractures. This is a time when services are often cut back so it is important to fight to maintain input. It is important to be quite vocal in explaining that this is a critical period for maintaining mobility.

Try to ensure that someone is monitoring the person with Rett syndrome's back (particularly if there is already a scoliosis) and hips, usually by doing check X-rays. Do remember, however, that some people with Rett syndrome develop more severe physical problems than others, whilst others are fine with a minimum of input.

## **Screaming episodes**

This is one of the commonest and most distressing problems through adolescence and into adulthood. A proportion of people with Rett syndrome develop screaming episodes by day or sometimes by night, which can go on for hours at a stretch. Sometimes they may have outbursts of laughter.

Parents and carers often feel quite powerless to deal with these, and it is very upsetting when the people with Rett syndrome appear to be unhappy or in pain. (See Screaming Episodes factsheet for recommendations).

## **Transitional care/Continuity of Services**

Adolescence is a time when many aspects of the person with Rett syndrome's physical and emotional health requires attention, but also a time when she or he is often moving from school and from paediatric services. Try to ensure that education and social services have good transitional care plans, and that the person with Rett syndrome's therapy input will be maintained into adulthood. Try to ensure that there is continuity of medical services when they leave the care of a paediatrician. Good review assessments before these handovers take place are essential.