



RETT SYNDROME ASSOCIATION UK

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Comprehensive Health Care in Rett Syndrome: a check list

Some health points which may need particular attention in people with Rett Syndrome

A. Kerr April 2002, updated March 2004

Health Area	Research Shows	Action Points
Sleep	Day sleep and night waking is common	establish day/night routine as toddler
Vision	Is particularly good	Check in case spectacles are required
Hearing	Is particularly good	Check occasionally for wax
Teeth	Are generally good	Regular checks, avoid sweet drinks
Feeding	Problems in retaining & chewing	Advice on feeding techniques may help
Nutrition	50% more calories needed than expected	request advice from dietitian
'Heartburn'	Reflux of stomach acid is common	good posture, antacids, rarely surgery
Lot of stomach air	Common, studies in progress	no cure so far, go for warmth and comfort
Constipation	common in all profound disability	good diet, lots of drinks, active lifestyle
Speech	the brain is poorly equipped for this	do encourage, a little speech is possible
Other communication	intuitive relating very good	two way musical communication is valuable
Voluntary hand use	the brain is poorly equipped for this	Encourage and expect some progress
Spontaneous movement	the brain is well equipped for this	best in music, dance, swimming, riding
Deep fast breathing	Hyperventilation, driven by brain	ignore, allow quietness if distressed
Breath holding	is driven by brain	ignore, allow quietness if distressed
Vacant spells	Not usually epilepsy, often apnoea	avoid unnecessary anticonvulsants
Seizures	(epilepsy) about 45% but less later	anticonvulsants may control these
Agitation	a fundamental Rett problem	accept it, allow quietness if distressed
Position sense	Reduced awareness of position	physiotherapy can help to adjust this
Muscle tone	Floppy at first, tight later	encourage active & varied physical activity
Posture	Poor because of last three items	good support for seating, standing and moving
Joint deformities	Apt to occur because last four items	regular exercise, ankle splints, rarely surgery
Scoliosis	The back affected like other	active movement, surgery if increasing

	joints	fast
Fractures	Inactivity weakens the bones	active movements strengthens bones
Walking	is possible for most but seldom easy	experience in movements using the whole body
Cold blue feet	Sympathetic tone is high due to poor autonomic restraint	treat skin gently, encourage activities using whole body
Toileting	Habit training is possible	develop a good routine, provide a supported set

Make sure your daughter attends 'well women' clinics and breast screening. Look out for common causes of pain and discomfort - ear infections, athlete's foot (tinea), vaginitis, urinary infection, dental abscess and so on. People with Rett syndrome do carry some extra risk of dying early because of their many problems and extreme dependence but many will outlive their parents. They always need a lot of support and therapy. No one person can carry the physical burden of care indefinitely or alone. If you don't look after yourself you won't be there to look after her. It is a good for her to know and be known to more than one family circle. Future care by other people will be better if you choose and influence it from the beginning. So do accept help, from school and day centre and respite through family, friends, foster-friends or care centre. Be prepared to plan ahead and select a small residence (4-6 people) for your daughter's later years, near your home so that you can easily spend time there too. She will always need the love of her family.