Having a Baby

Restricted Growth Association

Working to benefit people with restricted growth and their families

HAVING A BABY

This booklet is written primarily for prospective parents where one or both is of short stature. It seeks to answer many of the questions people have about pregnancy and childbirth, including:

- What is the risk of my passing my condition on to my baby?
- Will my restricted growth condition affect my chances of getting pregnant?
- Will pregnancy and childbirth be dangerous to my health?
- What tests are available to find out about my baby's condition?
- What can I do if I'm finding it hard to get the help and advice I need?
- How will my baby be delivered?

Taking the decision

Maternal and paternal instincts – the desire to bring new life into the world – are some of the strongest urges we humans feel. For anyone responding to this urge, many factors affect the decision about whether or not to have a child: Are we ready for the responsibility of bringing up children? Is this what we want at this stage of our relationship? Can we afford financially to have children? For people with restricted growth, there are additional issues to think about. These include:

- the physical demands it puts on the mother, where she has a restricted growth condition
- what the likelihood is of passing on a restricted growth condition
- your feelings about the possibility of having a baby with a restricted growth condition
- the possibility of having a severely, or even fatally, disabled baby, such as one born with a double dose of achondroplasia (see 'Genetic counselling' on the next page).

Physically, there is no reason why most women of restricted growth should not be able to have a healthy baby. Most have normal fertility and can safely carry a pregnancy through to delivery.

Some women do come up against prejudice in their contacts with health professionals and others who think that, for some reason, people of restricted growth should not have children.

"The gynaecologist I first saw at the hospital was extremely rude. He made lots of nasty comments about my height, such as "Oh, you're not very tall – you're actually very small, aren't you." He even made a point of asking me how I would cope if I had a child of normal stature. He said: "When it reaches 6 or 7, it will be taller than you and will look down on its parents." I looked him straight in the eye and said, "It doesn't matter whether the baby is tall or small, it will always look up to its parents, because that's the way we will bring our children up."

People's prejudices may be disguised as 'genuine concerns', e.g. about the physical rigours of pregnancy and the effects on someone of short stature. However, what they are really expressing is their own lack of understanding of restricted growth and their inability to see people who are 'different' as full human beings.

> *It's okay to be small and pregnant. That's it. No argument. No one has the right to tell you otherwise. If they try to, don't believe them. They're trying to deprive you of your own rights and the joy of having a child.*

Genetic counselling

Where one or both parents have a restricted growth condition that has a genetic cause, there is a risk of passing on the condition. What prospective parents need above all is information in order to understand these risks. Many couples seek *genetic counselling*, where they can find out this information. Genetic counselling is important both to people who want information before making their decision as well as to those who have already made their decision to have children, but want to understand the possibilities of passing on a condition.

Genetic counselling involves confirming the diagnosis of the parents' conditions (or condition, if only parent is of short stature), discussing how the gene that caused the restricted growth condition is inherited and calculating the risk of passing on the condition to children. Where both parents have *different* restricted growth conditions, genetic counselling is necessary to determine the risks of their children being affected by either or both of their parents' conditions.

The risks depend on the inheritance characteristics of the different conditions. With restricted growth conditions, there are three main types of inheritance:

- **Dominant Inheritance**

  For conditions such as achondroplasia, pseudoachondroplasia, hypochondroplasia and SED that are inherited in this way, only one copy of the gene that causes the condition is necessary for an individual to have that condition. If one or both parents have restricted growth, this gene could come from either of them or the child could be unaffected. If both parents have the same condition and the child inherits the gene from both of them, they will have an extreme form of the condition which may not allow the child to live beyond birth.

- **Recessive Inheritance**

  For conditions such as diastrophic dysplasia, 3M syndrome, Jarco Levin syndrome and cartilage hair hypoplasia that show recessive inheritance, two copies of the gene that causes the condition are needed, one from each parent, for the child to show the condition. The parents themselves will only have the condition if they have two copies of the gene. If two parents with the same recessive condition have a child, the child will always have the condition as well. If only one parent has the condition, it is extremely unlikely that their child will have the condition but they will be a carrier of the condition.

- **X-Linked Inheritance**

  Several genetic conditions, such as SED tarda and some forms of chondroplasia punctata, are caused by genes carried on the X-chromosome (one of the two types of sex chromosome).

  Men with the condition carry one affected X-chromosome and one (unaffected) Y-chromosome. Women who carry the affected X-chromosome will always have one unaffected X-chromosome and do not, therefore, usually show features of the condition, but they can pass it on to their male children.

Becoming the expert

There is still some ignorance among some health professionals about the genetics of restricted growth conditions. The RGA is seeking to address this through its **What is...?** series of booklets which contain information about

genetics. As well as answering some of your own questions, you can use them to inform professionals caring for you by passing copies of the relevant booklets on to them.

> *When I was pregnant, the consultant told me: "Normal-sized husband, normal-sized baby" and since my husband is of average stature, I believed I was going to have an average-sized baby. The consultant was simply wrong, which meant rethinking everything when my baby arrived.*
> *Mother with achondroplasia*

Although many paediatricians and some well-informed GPs are able to give genetic counselling to patients under their care, bone disorders are a complex group and it usually wise for genetic counselling to to be given in a centre with experience of these disorders. Your GP should be able to refer you on to your local Regional Genetic Centre (RGA has a list of these in its main office), although some GPs may not be aware of the availability of genetic counselling services. You may need to ask to be referred.

> *During my stay in hospital I spoke to many of the staff about achondroplasia as they had never come across this condition before. The hospital now has my name and details on file in case they ever have another mother who has a baby with achondroplasia and who needs help and advice.*

Making choices

The issue of genetics raises lots of questions about individual rights and choices, and touches deeply-felt personal, moral and religious beliefs. Tests may identify abnormalities in the child at a stage when termination of the pregnancy is possible – allowed by law up to 24 weeks. For example, if both parents have achondroplasia, there is a 1 in 4 (or 25%) chance that the baby will inherit the achondroplasia gene from both parents (the so-called 'double-dose'). Where this is the case, the baby will not survive beyond birth. Tests can be carried out on the foetus to determine whether it has this condition, in which case the mother will be offered – or even encouraged to have – a termination. This can be a very difficult and stressful time for both mother and father, and is something that they need to think about and discuss in advance.

Before becoming pregnant, my partner and I had genetic counselling, which involved a chat with a Genetic Nurse who explained the odds of what condition the baby might be born with – achondroplasia, double-dose or average stature. This was very important as it gave us an understanding about the risks of double-dose. It helped us prepare ourselves for the possibilty and what steps we would take if that happened. I would strongly recommend anyone in our situation to talk to a Genetic Nurse beforehand.

We didn't tell anyone about the pregnancy until I'd had the tests and we'd got through those first three months. We'd decided on a termination if the baby was double-dose and in any case, lots of women have miscarriages during the early weeks. It was really hard – I didn't dare believe everything would be okay. We didn't want to raise anyone's hopes, especially our own and our families'. It was only when the danger time was past that I allowed myself to relax, enjoy being pregnant and look forward to being a mother.

Genetic counselling gives couples the information to make their own decisions about the best course of action to take. At this point the decisions about the pregnancy can only be made according to your individual beliefs and conscience. Some women and their partners may make the choice to refuse the offer of tests and let the pregnancy take its course.

I strongly believe that abortion is wrong and so rejected the option of a test. Fortunately, my child was born healthy. He has achondroplasia like me and we feel blessed to have such a beautiful child.

Others may make the difficult decision not to have children.

After much agonising my husband and I decided not to have children. We did not want the risk of passing on my condition. Also, I've had so many medical problems over the years. I couldn't tell how I would cope.

People who decide not to have children of their own can still experience the joys of parenthood, by adopting or fostering children in need of a loving home. It is a sad fact that some children of restricted growth are rejected by their parents, who cannot come to terms with their child's condition or feel unable to cope with a child who is different and decide to have their child adopted. Other circumstances, such as family breakdown, parental

illness or death, may result in a child of short stature being put up for fostering or adoption.

Couples where one or both partners is of restricted growth often feel that through their own experience they have a great deal to offer such children. The Restricted Growth Association has produced a separate booklet in the **Lifestyles** series called *Adoption*.

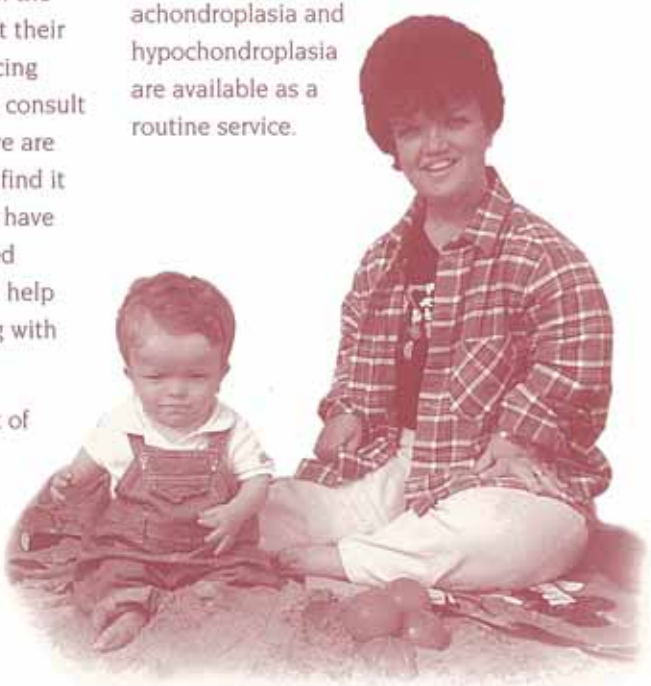
Conception

Some people of short stature are concerned that their restricted growth condition will affect their ability to conceive. For the vast majority of men and women the condition itself *will not* affect their fertility. If you are experiencing difficulties with conceiving, consult your doctor for advice. There are many reasons why couples find it hard to conceive and these have nothing to do with restricted growth. Your doctor should help you explore ways of dealing with the situation.

A few people do find the act of intercourse difficult due to physical constraints with themselves and/or their partners. This is particularly true for people with deforming processes, such as diastrophic dysplasia. Some women of restricted growth have a smaller-than-average vagina which may make penetration difficult or painful. Vaginal dilators can be used to make intercourse more comfortable – and again your doctor should be able to advise you. Also, in some short-statured women, the cervix is very far forward (anterior), but this is not thought to affect fertility.

Prenatal diagnosis

Once pregnant, you will have to consider the question of prenatal tests. At present, only tests for achondroplasia and hypochondroplasia are available as a routine service.



With other conditions, tests are still only at a research stage – if indeed they will ever be possible at all. This makes accurate assessment of the genetic risks by a genetic counsellor all the more important.

Methods used for prenatal diagnosis include the non-invasive ultrasound scan, and amniocentesis or chorionic villus biopsy test where the foetal tissue is investigated.

- The ultrasound scan produces an image of the foetus and some major abnormalities can be seen as early as the 16th week of pregnancy.
- Amniocentesis involves taking fluid from the womb, and foetal cells in this fluid can be cultured and examined for chromosome abnormalities; or the build up of a chemical in the fluid can be measured which reveals an enzyme abnormality in the foetus, e.g. Morquio's disease.
- The chorionic villus test involves taking a piece of tissue from the developing placenta, which can then be examined in a similar way to the amniotic fluid. The advantage of the chorionic villus test is that it is carried out at around the 9th week of pregnancy, whereas the amniocentesis test is usually not done for a further month. However, there is thought to be a slight risk of miscarriage following these procedures of between two and four per thousand pregnancies tested (0.2–0.4%).

"During my pregnancy, I had about five scans and was monitored quite closely. When I was about ten and a half weeks pregnant, I was told about a genetic test that could be done. It involved taking a DNA sample from the unborn baby to identify what condition it had and whether it might be double-dose. Even though there is a very slight chance of having a miscarriage as a result of having the test, we would still go through it again if we decided to have another baby."

Physical demands of pregnancy

All women when embarking on a pregnancy will be anxious about possible difficulties that lie ahead,* and this will be especially true for women of restricted growth.

One of the main factors in pregnancy is that baby is large in relation to size of the mother.

This booklet was written by Hugh Hillyard-Parker, with the help of the many members and their families who have contributed their experiences towards it. Their contributions were compiled by RGA Information Officer, Ros Smith.

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The RGA encourages members to share information and sources of help.

Other RGA publications

What is...?

The **What is...?** series looks at particular restricted growth conditions or issues. The titles in this series are:

Achondroplasia

Hypochondroplasia

Pseudoachondroplasia

SED

Diastrophic dysplasia

Rarer syndromes

Lifestyles

The **Lifestyles** series focuses on particular aspects of living with a restricted growth condition. The other titles in this series are:

Babies and the Early Years

Going to School

The Teenage Years

Careers, Employment and Education

Adoption

The Later Years

Driving a Car

Bikes, Trikes and Ride-on Toys

Sporting Opportunities

What Shall I Wear?

Limb Surgery

Personal Hygiene

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