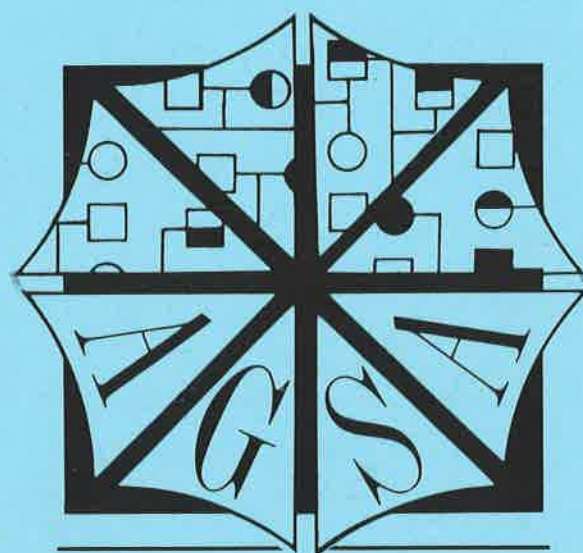


A G S A

NEWSLETTER

JUNE 1990

ISSUE NO. 4



THE • ASSOCIATION
OF • GENETIC • SUPPORT
OF • AUSTRALASIA

ISSN 1033 - 8624

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C/- 44 Rawson Street
EPPING NSW 2121
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The views expressed in this Newsletter are not necessarily those of AGSA

Share a Thought

We make no apologies for reprinting this article from the Down's Syndrome Association's Spring edition of *NEWS* for it struck a chord with us! It is an extract from an address to the USA National Down's Syndrome Congress by Diane Crutcher, the Executive Director:

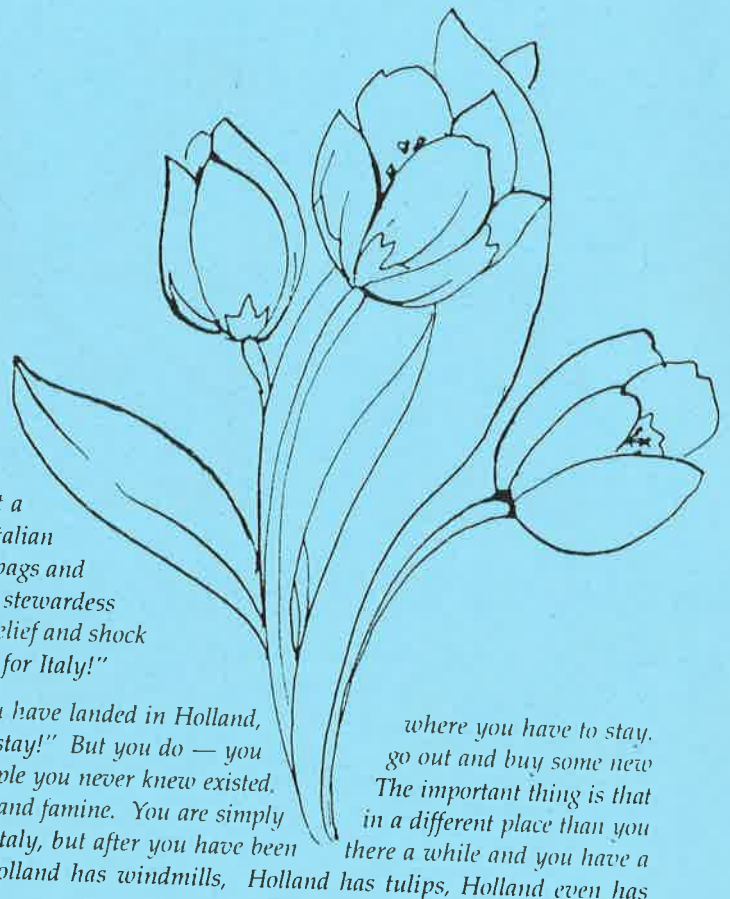
"When you are going to have a baby, it is like you are planning a vacation in Italy, you are all excited — you get a whole bunch of guide-books and you learn a few phrases in Italian so you can get around. When it comes time, you pack your bags and head for the airport for Italy. Only when you land and the stewardess says: "Welcome to Holland" you look at one another in disbelief and shock and say: "Holland? What are you taking about? I booked for Italy!"

Then they explain there has been a change of plans and you have landed in Holland, "But I don't know anything about Holland! I don't want to stay!" But you do — you go out and buy some new guide-books. You learn some new phrases, and you meet people you never knew existed, you are not in a filthy, plague-infested slum full of pestilence and famine. You are simply in a different place than you planned. It is slower-paced than Italy and less flashy than Italy, but after you have been there a while and you have a chance to catch your breath, you begin to discover that Holland has windmills, Holland has tulips, Holland even has Rembrandts!

Of course, everyone else you know is busy coming and going to Italy. They are all bragging about what a great time they had there, and for the rest of your life you will say "Yes. That's where I was going. That's what I planned!" The pain of that will never, ever go away. You have to accept that pain because the loss of that dream, the loss of that plan, is a very, very significant loss. But if you spend your life mourning the fact that you did not get to Italy, you will never be free to enjoy the very special, the very lovely things about Holland."

Many thanks to Diane Crutcher for sharing these thoughts. (Editor) — Contact A Family

Many thanks also to Down's Syndrome Association and Contact A Family. Amazing how things get around!
(Editor — AGSA)



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- CONTACT CORNER
- SUPPORT GROUP NEWS BULLETIN
- RESOURCES AND AIDS
- PROFILE — A-Z GENETIC CONDITIONS
(Insert — *Please remove if required for your files*)
 1. Turner Syndrome
 2. Achondroplasia

EDITORIAL

In the "Editorial" (March '90) mention was made of the pursuit of EDUCATION and AWARENESS of genetic disorders - the need for professionals and lay persons to walk hand-in-hand.

What do we actually mean:

- | | |
|-----------|--|
| education | the act of process of acquiring knowledge |
| | the act of process of imparting knowledge |
| | a particular kind of instruction or training |
| awareness | having knowledge |
| | informed of current developments |

Key Words LEARNING/REASONING/AWARENESS

AGSA aims to:

- educate the medical and allied health professionals and the community about genetic disorders

In keeping with the foregoing, EDUCATION and AWARENESS have been at the forefront of the community over recent months.

AWARENESS WEEKS have been held for:

- Muscular Dystrophy
- Neurofibromatosis
- Down Syndrome and
- Respite Care (*see Letters to the Editor*)

Coming shortly - National Diabetes Awareness Week
"Living A Healthy Lifestyle"
July 15-21, 1990

In the area of EDUCATION

- * Trainee Genetic Counsellors Orientation
Workshop/Seminar
30th-31st May, 1990

run by NSW Genetics Education Program at which
Support Group Services were addressed.

EDUCATION - and YOU Seminar
2nd June, 1990

An overview of the education options
available to families.

*(detailed report further through Newsletter)

In addressing some of these issues we are hopefully increasing YOUR knowledge. However, the Government do not perceive education as being worthy of consideration.

Where do we go now?

(*See President's Report*).

* * * * *

PRESIDENT'S REPORT

21st June, 1990

Response to our newsletters has been overwhelming and I am grateful to hear so many people speak so highly of Ros Smith's contributions to AGSA, and in particular her efforts in producing what is obviously a widely accepted newsletter. The newsletter is responsible for attracting new members which is even more pleasing.

In my last letter to you I talked positively about a pending approval for taxation deductibility for donations. After considerable negotiations with the Taxation Department they have chosen at this stage to reject the AGSA application. This is a most serious set back and if it cannot be resolved, places AGSA's future at serious risk. Our application was first lodged in September last year, nine months later the Federal Government has chosen to reject our overtures. Very clearly Federal politicians have little understanding of the issues with which we are confronted, and I find it even more galling given that AGSA is really doing what, in my view, is the responsibility of the Federal Government.

I have no intention of taking this decision laying down and have retained a highly respected tax accountant, who has generously agreed to resume discussions with the Commissioner of Taxation on our behalf, at no expense. This is not the first time that professional people have offered their help to AGSA free of charge and to me this reflects the regard that we are held in from some sections of the community. It is indeed a pity that politicians and bureaucrats cannot extend to us the same courtesy.

Aside from the assistance that is being offered by this consultant, I would respectfully ask the President's of all affiliated support groups to write to me and offer their support. Your letters will accompany a further submission by me to the Federal Treasurer. I would also appreciate the support of those health professionals who were very much behind the establishment of AGSA and who see us as their partners with a common goal.

I believe that AGSA has a very important role to play in the community and we must utilise every resource available to us to achieve tax deductibility.

Whilst the Federal Treasurer has indicated that there may be some room to move in the areas of research and respite care, he has unequivocally rejected a request for deductions for donations for the purposes of education. AGSA sees one of its many priorities as education of the community at large and this really is an absolute must.

I do not believe that we will ever obtain the financial support we need if tax deductibility is not available for donations.

I can assure you as your President I will continue to work energetically and do not intend taking 'no' as an answer. Your continued support would be greatly appreciated.

R J M MacDiarmid
President

CLASSIFIEDS

FOR THE PRESENT TIME
THE CONTACT NUMBER WILL BE
(02) 868 2559.

* * * * *

RATES FOR THE CLASSIFIEDS

Full page — \$160
1/2 page — \$90
1/3 page — \$60
1/4 page — \$50
1/6 page — \$35

Artwork and typesetting for classifieds
additional charge

* * * * *

INTERNATIONAL NEWS

6th Annual Postgraduate Symposium National Fragile X Foundation u.s.a.

"Diagnosis and Management in the 90's"

Saturday-Sunday — 15th-16th September, 1990
Westin Hotel, Washington. D.C.
Registration by 24th August, 1990

To: Cheryl Richardson,
Genetics and IVF Institute
3020 Javier Road
FAIRFAX, VIRGINIA - 22031
Telephone: 703 698 3948

* * * * *

Mrs Lesley Green, Hon. Secretary
Research Trust for Metabolic Diseases in Children
(RTMDC)
53 Beam Street
NANTWICH, CHESHIRE. CW5 5NF. ENGLAND

RTMDC was founded in November, 1981 after Lesley and Peter Greene discovered their first child was suffering from 'a rare, incurable and inherited metabolic disease' called Cystinosis. They decided there must be many families receiving similar news for this, and other metabolic diseases, and that a research trust could offer financial support to help scientists and emotional support to help families. The concept of an umbrella organisation for inborn errors of metabolism was the first of its kind in the world. RTMDC now has an international reputation and both Lesley and Peter remain actively involved in its progress and future development.

9th Parents' Conference of RTMDC
Saturday, 15th September, 1990

Editors Comment:

I will be attending this Conference if any one wishes me to follow up any concerns they may have regarding their own child's metabolic condition.

CONTACT-A-FAMILY

WHAT CONTACT-A-FAMILY DOES

Contact-a-Family is a national charity which supports families who have children with different disabilities and special needs. Families with a disabled child often feel isolated and Contact-a-Family brings them together through mutual support and self-help groups.

A quarterly newsletter, Share an Idea, is circulated to a wide range of parents and professionals and provides information and advice on a range of topics. Share an Idea is available quarterly, on subscription (£5.00) per annum.

For further information about Contact-a-Family and its services, please write to:

The Information Department
Contact-a-Family
16 Strutton Ground
LONDON SW1P 2HP
Telephone: (071) 222 2695

"I think it would be a lovely idea if an article on Contact-a-Family could be published in one of your Newsletters and indeed we would be pleased to reciprocate. I hope you will find the enclosed suitable. From time to time we publish requests for contact for individual parents in Share-an-Idea (our Newsletter) and should be very pleased to hear of any suitable links at your end."

Christine Lavery
National Development Officer
for Rare Handicap Groups

* * * * *

CONTACT-A-FAMILY

Being told that your child has special needs and disabilities is probably one of the most painful experiences that a parent may live through. Many parents in this situation, while having early contact with many medical professionals, also feel a great need to talk with other parents who may be in a similar situation.

According to the recent government-commissioned OPCS (Office of Population and Censuses Survey) statistics, children having special needs and disabilities in the nought to 16 years age group now number 360,000. Of these children 355,000 live at home with their families, where every member will be affected in some way by the caring needs and caring tasks for that child.

Contact-a-Family is a national charity whose primary constituency is those families in the UK whose children are diagnosed as having special needs and disabilities. It works nationally with a network of parent support/mutual aid groups, which fall into two types.

- * Approximately 600 local groups run for parents of children with a broad range of special needs.
- * Over 200 national groups run by and for parents of children diagnosed as having a rare specific disability or syndrome.

Many parents belong to both types of group in an effort to address the need for mutual support and knowledge.

Both these types of groups are supported within Contact-a-Family by development officers who can offer help and advice on setting up, maintaining and developing these groups. Many of the nationally run groups have started from small beginnings: one or two parents and the support of Contact-a-Family, plus a great deal of enthusiasm from the pioneering group leaders, have successfully become well run supportive groups.

Contact-a-Family run an information advice and linking service for parents and professionals across the country. Two specialist parent advisers are available and link mainly to known support groups, but will link to individuals if more appropriate.

Many parents and professionals wrongly assume that because the diagnosis may be unusual a support group does not exist.

A call to Contact-a-Family can not only answer this query but also give you detailed information about the group's progress and just what parents can expect from the group.

Being involved with setting up these groups means we would also be involved with some groups in the very early stages.

There is an optional membership and affiliation scheme for these groups, plus a subscription service for any individuals who may wish to receive the quarterly newsletter, Share-an-Idea.

Two development officers will also offer support to any parent or professional interested in starting such a group.

LETTERS TO THE EDITOR

Mrs Jenny Rollo
Cornelia de Lange
Syndrome Support Group
135 Princes Street
Putney NSW 2112

Mrs Roslyn Smith
AGSA
44 Rawson Street
Epping NSW 2121

Re: "EDUCATION AND YOU" Seminar 2nd June, 1990

Dear Ros

Thank you very much to the Family Education Unit (North Ryde), and to AGSA for running the "Education and You" seminar in June.

As I am perfectly happy with my son's placement at "Special" School, my attendance at the seminar was really to keep up to date with education trends in NSW. The speakers adequately fulfilled that need.

I was particularly interested to learn of the differing needs of so many other families, and I now feel more in sympathy with the need for integration into public schools.

The bonus was meeting old and new friends! I would like to recommend that other parents take the time to attend these functions, as so much can be gained. I look forward to the next seminar.

Yours sincerely

Jenny Rollo

A "Thank You" card was also received from the Johnson family requesting their appreciation be passed on to the Education and You organising Committee, child care workers and for the use of 44 Rawson Street as the creche.

Editors Comment:

I was delighted to offer my home and more importantly that parents (both parents) were able to attend without worrying about their children.

* * * * *

Food for Thought

Victoria
6th June, 1990

Dear Sir/Mesdames

This being Respite Care Awareness Week, it is disturbing indeed to feel the need to write this letter - addressing the Committee of Management on the recent closure of the Philipdale Court Respite Care House.

It is becoming increasingly obvious to those of us with children and young people with special needs, multiple handicaps and severe intellectual disabilities, that we are not being catered for within the new education and 'life' programmes. And ALSO in the area of Respite Care.

The most suitable respite care offered, in fact, the ONLY suitable respite care offered, to those of us with children depending on familiarity, and good loving individual understanding of needs, has been that provided by Knoxbrooke Committee of Management, and more recently at Philipdale Court. As far as I am concerned, Philipdale Court has been a prime, and superb example of provision for care, with the emphasis on 'special needs' children and young people.

The closure of the Knoxbrooke facility leaves several families without respite — and with the on-going worry of 'emergencies'. I feel that consultation with these parents regarding difficulties experienced in maintaining the service, may have prevented — or may yet prevent the permanent closure of Philipdale Court and the loss of an excellent staff member.

I urge the Committee to be aware of the needs of all families within the Knoxbrooke community, and increase communication with those it represents.

I remain

Sincerely

Name and address
supplied

The above is not an isolated incident — unfortunately this is happening all too often.

Editor

* * * * *

URGENT LETTER WRITING REQUIRED NOW!!

A considerable number of children wear hearing aids. Most aids would have been supplied by the National Acoustic Laboratories (NAL), free of charge: (Remember signing a form to lease them from the Federal Government?).

It has come to my attention via the North Shore Deaf Children's Association, (of which I am a member), that the Federal Government proposes to terminate the services of the National Acoustic Laboratories, throughout Australia, in favour of the Private Sector.

This efficient government agency caters for 65% of the people wearing hearing aids in Australia. These include children to 21 years of age and means tested pensioners. Many of the tests advised by NAL are recognised and used world wide. NAL is also concerned with research and industrial noise. While children are a small percentage of

the 65% of the hearing impaired NAL service, they devote a large portion of their time and budget to the children. NAL employs trained audiologists to access the hearing loss and to fit the aids, which is not an easy task where children are concerned and entails a lot of time. Such consumption of time the private sector would find non-profitable, and I fear that they would serve this group very badly. I understand that audiologists are few and far between in the private sector. Apparently any untrained person can sell you a hearing aid.

In the USA many children wear only one aid because it is impossible for their parents to afford the second aid from their private system, and thus the quality of life for those hearing impaired children is lowered greatly.

Hearing impaired children are disadvantaged enough already without suffering further for the sake of the private sector making more profit. This is one area where government staff are doing a very good job — let us make sure that they are allowed to continue.

On 28th May The Industries Assistance Commission conducted a hearing, held at Goulburn Street, Sydney, at which the NSDCA made a submission opposing the proposal to disband NAL. Apparently the existence of this IAC hearing was well known, in advance, by suppliers and retailers of Hearing Aids, but few of the support groups for the hearing impaired, knew about the hearing until 10 days before.

I feel that it is imperative that all parents of hearing impaired children should voice their disapproval of the Federal Government's proposal to terminate the National Acoustic Laboratories, by writing letters to Federal Politicians. Listed below are names and addresses. Please write these letters now. The matter is urgent. Also please advise any hearing impaired pensioners you know of, to do the same. In this type of battle, numbers count. A politician is more likely to notice thousands of individual letters voicing the same opinion, no matter how simply written than a few perfectly drafted and typed submissions.

Thanking you in anticipation of your efforts.

Isabel Glasson

Hon Mr R J L Hawke ACMP
Prime Minister
Parliament House
CANBERRA ACT 2600

Dear Mr Hawke

* * * * *

Hon P J Keating MP
Deputy Prime Minister and Treasurer
Suite 17, Capital Centre
41-45 Rickard Road
BANKSTOWN NSW 2200

Or Parliament House

Dear Mr Keating

* * * * *

Hon Mr B L Howe MP
Minister for Community Services and Health and
Minister Assisting the Prime Minister for Social Justice
Parliament House
CANBERRA ACT 2600

Dear Mr Howe

* * * * *

Senator The Hon J N Button
Minister for Industry, Technology and Commerce
4 Treasury Place
MELBOURNE VIC 3000

Or Parliament House

Dear Senator Button

* * * * *

Your own Federal Member

OR

Parliament House
CANBERRA ACT 2600

* * * * *

ROYAL HOSPITAL FOR WOMEN

UNDER THE AUSPICES OF THE BENEVOLENT SOCIETY OF NEW SOUTH WALES



188 OXFORD STREET • PADDINGTON • N.S.W. • 2021
TELEPHONE: 339 4111

PLEASE QUOTE

13th June, 1990

The Editor

The Association of Genetic Support of Australasia
44 Rawson Street
EPPING 2121

Dear Sir/Madam

Re: Adoption and Genetic Disease

It could be helpful to your members to know of services available in NSW, and most other States, where a genetic disease is diagnosed.

If an adopted person develops such a disease and the treating medical specialist believes either diagnosis, treatment or prognosis would be assisted by detailed information from the natural family or that the natural parents should be informed of the disease, the Adoption Agency involved or the Department of Family and Community Services (Family Information Section, PO Box 3485, Parramatta, 2124) will attempt to trace the natural mother and obtain the information needed.

If a parent (who has in the past relinquished a child for adoption) develops a genetic disease, or if subsequent children do, and if his/her medical specialist believes that the adopting parents or the adopted adult should be informed of the presence and nature of the disease, then either the Department or the private adoption agency which placed the child, will agree to attempt to trace and inform the adoptive parents or adult adoptee.

Clearly such tracing is a very sensitive and difficult process, but can be extremely helpful, and informative for all parties to adoption.

I would be happy to answer queries about this process, on telephone (02) 339 411 or queries could be addressed to the Family Information Section referred to above [telephone: (02) 683 9999].

Yours sincerely
(Mrs) Petrina Slaytor
Social Worker

FIRST LYING IN HOSPITAL IN NEW SOUTH WALES

* * * * *

NEWSLETTER DEADLINE FOR SEPTEMBER 1990

The deadline for information for the June issue of the AGSA Newsletter is **31st August, 1990**. If you wish to publish an article or item of interest in the September Newsletter, please forward relevant information to:

Mrs R Smith
44 Rawson Street, EPPING 2121

GENETIC SERVICES

REPORT FROM TRAINEE GENETIC COUNSELLORS' ORIENTATION WORKSHOP/SEMINAR

The NSW Genetic Education Program organised an orientation workshop/seminar for NSW Genetic Counsellors at the request of the NSW Genetic Services Advisory Committee. The seminar was held at Royal North Shore Hospital, 30th-31st May, and attended by 16 Counsellors many of whom were recent appointments.

The seminar allowed much needed opportunities for counsellors, especially those who are geographically and professionally isolated, to meet and discuss common interests and concerns. The sharing of ideas knowledge and skills, was seen as very beneficial by all those who attended. The seminar included lectures on the History and Ethics of Genetic Counselling, Prenatal Testing, Genetic Education Resources, Cytogenetics, Counselling issues, Outreach Services and a well received section on Support Groups and the Family's Perspective.

Ros Smith spoke as a parent, support group member and Honorary Executive Director of AGSA. The advantages of a good partnership between parents, support groups and health professionals was emphasized and is already much in evidence in the increasing use of AGSA by Genetic Counsellors and others working in related fields.

Counsellors are planning regular professional meetings in the future and have shown a particular interest in further input from families in order to continually develop the skills and sensitivity to meet the needs of affected families.

AGSA members may be interested in the following list of names and addresses of Genetic Counsellors employed in NSW:

Ms Miriam Briet (02) 438 7280
Genetic Counsellor
MSAFP Program
(Maternal Serum Alpha-Fetoprotein)
C/- Ground Floor, Maternity
Royal North Shore Hospital
Pacific Highway
ST LEONARDS NSW 2065

Ms Lynn Rawlings (02) 438 7280
Genetic Counsellor
MSAFP Program
(Maternal Serum Alpha-Fetoprotein)
C/- Ground Floor, Maternity
Royal North Shore Hospital
Pacific Highway
ST LEONARDS NSW 2065

Ms Jan Roberts (049) 60 2206
Genetic Counsellor
Regional Medical Genetics Unit
Newcastle Western Suburbs Hospital
Turton Road
WARATAH NSW 2298

Ms Bronwyn Butler (02) 692 6273
Genetic Counsellor
Medical Genetics and Dysmorphology Unit
Royal Alexandra Hospital for Children
CAMPERDOWN NSW 2050

Ms Margot Latham (02) 692 6302
Genetic Counsellor
Medical Genetics and Dysmorphology Unit
Royal Alexandra Hospital for Children
CAMPERDOWN NSW 2050

Ms Gillian Brown (02) 339 4432
Trainee Genetic Counsellor
Royal Hospital for Women
Oxford Street
PADDINGTON NSW 2021

Ms Ann Colhoun (049) 68 1453 Ext 237
Prenatal Diagnostic Unit
PO Box 21
WARATAH NSW 2298

Ms Judith Elber (042) 29 3444
Genetic Counsellor
Wollongong Hospital
Crown Street
WOLLONGONG NSW 2500

Ms Kim Frumar (063) 33 1311
Genetic Counsellor
C/- Bathurst Base Hospital
BATHURST NSW 2795

Ms Sue Latham (049) 28 0940
Genetics Associate
Stockton Centre
STOCKTON NSW 2295

Ms Robyn Pedersen (02) 399 2156
Genetic Counsellor
Department of Medical Genetics
Hut J
Prince of Wales Children's Hospital
High Street
RANDWICK NSW 2031

Mr John Rae (067) 66 2555
Genetic Counsellor
PO Box 83
TAMWORTH NSW 2340

Ms Hazel Robinson (02) 399 2294
Genetic Counsellor
Fragile X Program
Hut J
Prince of Wales Children's Hospital
High Street
RANDWICK NSW 2031

Ms Meryl Smith (02) 633 6892
Genetic Counsellor
Department of Paediatrics
Westmead Hospital
WESTMEAD NSW 2145

Ms Lucille Stace (066) 231 1492
Genetic Counsellor
Lismore Base Hospital
LISMORE NSW 2480

Further information can be obtained by contacting the
NSW Genetic Education Program Co-ordinators:

Dr Kristine Barlow OR
Ms Amanda O'Reilly
PO Box 32
NORTHBRIDGE NSW 2063
Telephone: (02) 438 7324

MEDICAL ARTICLE

SHORT STATURE

THE CONDITION

Everyone is aware of the wide scope of heights of apparently normal school children of the same age. Fewer than three percent of normal children have heights more than two standard deviations below the mean for age. The heights of children with short stature due to some underlying disorder (pathological short stature) are all at or below the third percentile for age. A child with a height age which is less than two years below the chronological age almost always has pathological short stature, although this may not be true in late childhood as children are going through puberty at different ages. The height (length) age can be estimated as that age at which 50% of children have the same height (length) as the child in question. Children who are below the third percentile in height and in whom there is any question about growth rate, should be referred for a medical opinion.

There are many causes of pathological short stature. Two broad groups of children can be distinguished: those who have proportionate short stature and those who have disproportionate short stature, (ie short arms, legs, or a short trunk). The causes of proportionate short stature include hormonal disorders, chromosomal disorders (the chromosomes represent the packaging of genetic material), chronic illness in childhood, and gastrointestinal disorders. Disproportionate short stature usually indicates there is a malformation of specific bones (eg a limb deficiency), or a primary disorder of growth of the skeleton, technically known as a dysplasia, but in common usage called "dwarfing". Medical authorities in recent years have recommended the dropping of the word "dwarf", as it is a most imprecise term. At present, there are known to be over 150 distinct skeletal dysplasias. All are heritable, ie either inherited, or capable of being inherited.

PROBLEMS OF SHORT STATURE IN SCHOOL

Children with short stature have certain problems with schooling in common, but many problems which are individual. Common problems include:

- **Unsatisfactory Seating**

An inappropriate desk height or seat height may prevent the child from effectively completing school work.

- **Access**

Very small children, and particularly children with short stature and motor disabilities, may have problems with access to classrooms if there are many steps or if the door handles are too high.

- **Stigmatisation**

Both direct and indirect stigmatisation occur.

Indirect Stigmatisation occurs when social interaction and interpersonal interaction is not appropriate for the chronological age of the short statured child but more appropriate to

the height age, eg all adults find it very difficult to interact with a twelve year old with a height age of six years in a manner appropriate to the age of the child.

Direct Stigmatisation occurs inevitably with short statured children, eg name calling such as "dwarf", "midget". One effective way to counter this is to encourage the child to live with the name calling but respond to other children with his medical diagnosis eg child "Yes I am a dwarf but doctors don't use that word any more. They say that I have achondroplasia".

- **Utilisation of Amenities**

Toilets, toilet chains, washbasins, tuckshop counters, may all be too high for a very short child to use them effectively. A solution to each problem is usually self-evident. It requires some ingenuity by the parents and tolerance by schoolmates. A small stool to reach chairs and a little help from classmates go a long way to solving all these problems.

DRUGS USED IN TREATMENT

For many disorders leading to short stature, there is no specific drug therapy. For a few disorders there is hormone replacement therapy. However, it would be extremely unusual for medication to be taken at school for short stature alone.

SCHOOL PROBLEMS RELATED TO SPECIFIC FORMS OF SHORT STATURE AND CHILDHOOD

Each specific type of short stature in childhood will bring with it a distinct natural history and perhaps special problems with schooling. Three disorders need our attention because of their frequency and because they are prototypes for all other disorders leading to short stature. These are the Turner Syndrome, Achondroplasia and Spondyloepiphyseal Dysplasia (and Spondylo-dysplasias in general).

Turner Syndrome

Turner Syndrome is a chromosome deficiency found only in females. There is either a single X Chromosome or portion of one missing. The incidence of Turner Syndrome is 1 in 2,500 female births.

The main consequences of this are:

- short stature (average height 140cm)
- lack of secondary sexual characteristics
- infertility

There may be other medical problems related to Turner Syndrome. These are: eye, ear, heart, kidney and thyroid difficulties, sugar diabetes and high blood pressure.

Sometimes there are other physical features such as low set ears, low hairline, webbed neck, pigmented moles, bending out of the elbows and puffy hands and feet.

Apart from the short stature, these children may have difficulty with skills requiring normal space-form perception such as line-drawing and geometry. These difficulties can, however, in the main, be overcome with encouragement from teacher's and parent's and a concentrated effort on the part of the girls.

Depending on growth rate, many Turner Syndrome girls are now given growth hormone in their early

years. Hormone (oestrogen) replacement therapy is usually required at puberty to permit the development of female secondary sexual characteristics.

* A similar syndrome occurs in boys and girls where it is called Noonan Syndrome. In these children the chromosomes are normal.

Achondroplasia

These children show disproportionate short-limb short stature. Achondroplasia is the commonest of the readily recognized disorders leading to short stature. It is a heritable disorder, although many children are the only ones affected in their family.

Children with achondroplasia not only have short arms and legs but also have short fingers. Sitting height is generally in the normal range, so that seats and desks can be used appropriate to chronological age. However, a stool may be useful in the classroom and toilets.

There is an increased risk for fluctuating hearing loss due to glue ears in children with achondroplasia. Thus any suspected hearing loss should be taken seriously and brought to the attention of the parents so that they may seek a medical opinion. Squint (turned eye) occurs more commonly than usual. If it occurs, the parents should be informed, as it may only be manifest when the child is tired or concentrating. It should always be investigated by an ophthalmologist.

Several activities are relatively contraindicated. These include skate-board riding, roller skating, hula hooping and exercises which place undue stress on the low back.

Spondyloepiphyseal Dysplasia and Epiphyseal Dysplasias

The term "Spondyloepiphyseal dysplasia" simply means abnormal growth of the spine and ends of the tubular bones, eg arms and legs. As spinal growth is the most severely affected, these children have a degree of short-trunk disproportionate short stature. Seating is commonly a problem in the classroom. As in all children, good posture should be encouraged to prevent back strain.

Curvature of the spine scoliosis (= side to side curve) or kyphosis (front to back curve) may develop and require bracing or surgical treatment. Hands are usually of normal size and shape. Facial appearance is usually normal.

A high proportion of children with various spondylo-dysplasias have high-grade near-sightedness. This will require frequent supervision by an ophthalmologist with review at least yearly.

While there are very few complications in childhood, participation in body contact sports should be discouraged so as to reduce trauma to the back and joints. This recommendation also applies to children with epiphyseal dysplasia only. Water sports and team sports such as tennis and golf should be encouraged.

EDUCATIONAL IMPLICATIONS

Short stature is not associated with intellectual disability. Certain groups of children with intellectual retardation, eg Down's syndrome, have mild concomitant short stature. For all other children with

short stature, difficulty with school work should motivate a search for other problems such as social immaturity, depression or special specific sensory problems, ie visual or hearing defects which are interfering with their school work. Remedial education will require dealing with specific underlying cause(s) of school failure.

FURTHER INFORMATION

Professional information can be obtained from a child's usual doctor in New South Wales or by contacting:

The Co-ordinator
Connective Tissue Dysplasia Clinic
Royal Alexandra Hospital for Children
CAMPERDOWN 2050
Telephone: (02) 692 6302

Similar Bone Dysplasia or Genetics Clinics provide these services in other states.

The organization **Little People's Association of Australia** exists to promote the exchange of ideas and information about coping with short stature. Its address is:

Little People's Association of Australia
The Summit Road
PORT MACQUARIE 2444
Telephone: (065) 82 0574

This association has produced a most useful monograph which all teachers of children with significant short stature should have. The monograph is titled **Information Guide on Persons of Short Stature**. It can be obtained through the Association.

For further information on Turner Syndrome, please contact:

Mrs Glenn Fisher
Turner Syndrome Association Inc
PO Box 112
FRENCHS FOREST 2086
Telephone: (02) 452 4196

Please Note:

Three small booklets on various aspects of Growth Hormone Treatment, written by the Australasian Paediatric Endocrine Group, sponsored by Pharmacia, are available.

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Minds are like parachutes —
they only function when open

PROFILE

A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile each Support Group/Disorder alphabetically thus increasing awareness within our membership of the range of genetic conditions. Also, it is hoped that where overlaps occur in conditions, Support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is 'Alive and Well' and happy to take referrals.

For your information we profile . . .

TURNER SYNDROME

AND

ACHONDROPLASIA

REMOVABLE INSERT
Please pull gently
from staple binding
to remove

TURNER SYNDROME

DIAGNOSIS

Approximately one-third of Turner individuals are diagnosed during the newborn period, one-third during childhood, and one-third during the late teens when it becomes apparent that they have failed to go through puberty.

A medical practitioner may suspect that a female has Turner Syndrome due to the presence of physical features associated with the disorder. A further and conclusive diagnosis can be made only by karyotyping (chromosomal analysis), — a precise means of identifying chromosome complement.

PHYSICAL CHARACTERISTICS

In addition to short stature, infertility, and the lack of secondary sexual characteristics, all of which are common to Turner Syndrome, the following is a list of additional physical characteristics that have been detected in Turner individuals. Most of these features are not exclusive to Turner. It is sometimes difficult to determine if a physical characteristic attributed to Turner is actually a result of the disorder itself, or the result of an independent inherited or environmental factor.

Nearly half of newborn Turner girls have puffy

hands and/or feet. These problems usually disappear in a few weeks or months so parents should not be too anxious about this. There may also be relatively pronounced skin folds in the neck which also disappear rather quickly. In some cases where there is a broad neck or more permanent skin folds (so-called webbing), on both sides of the neck, these folds, if they continued to be present and cosmetically disturbing, can be surgically removed by a plastic surgeon.

Approximately half of all girls with Turner Syndrome have problems during the first year of life with gulping and vomiting. In rare cases there may be a constriction of the connection between stomach and bowel.

Approximately 1 out of 10 girls with Turner Syndrome is born with a constriction of the aorta (main heart artery). This disorder, which is called coarctation of the aorta, is usually diagnosed during the first year of life or later in childhood. If the constriction is pronounced, it has to be surgically corrected. This can be done with very little risk, and leads to completely normal cardiac function. If the constriction is not present in the early childhood, there is no risk that it will appear later in life.

It should be noted that Turner individuals may not exhibit all of the listed features:

- Soft finger nails which turn up at the tips
- Low set ears
- Chronic middle ear infection (otitis media)
- Constriction or narrowing of the aorta (coarctation)
- Wide carrying angle of the elbows (cubitus valgus)
- Dysfunctions of the kidneys and urinary tract
- Folds of skin on inner edge of eye (epicanthal folds)
- Gastrointestinal and other feeding problems (during infancy)
- Heart murmur
- Reduced thyroid function (hypothyroidism)
- Low hairline
- Build-up of fluid in limbs during infancy (lymphedema)
- Short-sightedness (myopia)
- Other skeletal abnormalities
- Droopy eyelids
- Renal abnormalities (structural abnormalities and dysfunction of the kidneys and urinary tract)
- Broad chest with widely spaced nipples (Shield chest)
- Webbed neck

Other reported findings in Turner Syndrome are:

- Diabetes mellitus
- Dry skin
- High blood pressure
- Formation of scar tissue (keloid formation)
- Small jaw (micrognathia)
- Narrow high-arched plate (the top of the inside of the mouth)
- Pigmented moles
- Verbal/performance discrepancy

The three most common karyotypes of Turner Syndrome are:

Monosomy (Classic)

This classic form of Turner Syndrome is represented by the karyotype 45,X, meaning that all cells are missing an X-chromosome. This group generally exhibits more of the physical features (listed previously) than those who fall under other categories of the Turner Syndrome population. The incidence of monosomy Turner is about 50%.

Isochromosome

Normally, a chromosome divides longitudinally, but in the case of isochromosome it divides transversely. The result is a loss of all or a portion of one of the chromosome arms and the genetic material contained therein.

Two normal X-chromosomes are necessary for proper sexual development. The term, 'isochromosome', in reference to an X-chromosome with a normal long arm, karyotype 46,i(Xq), means that the short arm of the chromosome is missing and the long arm is duplicated. Women with this isochromosome are not likely to differ in appearance from monosomy individuals. However, they do have a slightly lower frequency of neck webbing and cardiovascular defects.

Isochromosomes account for a total of 12-20% of all Turner cases.

Mosaicism

When either the chromosome number or structure differs in different cells of an organism, the condition is described as chromosomal mosaicism.

One example of mosaicism in a Turner individual is represented by the karyotype 45,X/46,XX. In this variation only some of the cells are missing the second chromosome. These individuals tend to exhibit the fewest number of physical characteristics associated with Turner Syndrome. Up to 20% of this group are capable of menstruation, and there have been one hundred known pregnancies reported from women with this type of mosaicism. Those who do menstruate usually experience early menopause, beginning in the late twenties or early thirties.

Cellular combinations differ in mosaicism. For example, an individual may have some cells that are missing an entire X-chromosome, as well as some that are isochromosome, eg 45,X/46,X,i(Xq). Thirty to forty percent of all Turner Syndrome patients are mosaics.

Medical Management

Initial management requires referral to a medical geneticist to confirm diagnosis, clarify the exact chromosomal abnormality and obtain counselling about what can be expected in the future.

Medical Management should include regular visits to a Paediatric Endocrinologist who will monitor growth, thyroid function, blood pressure and administer the necessary hormone treatment for the development of puberty as well as any other endocrine related problems that may arise.

NB — Early diagnosis of Turner Syndrome is crucial if growth promoting therapy is to be started early enough to maximise final height. The consensus is that growth hormone treatment should be started as soon as there is evidence that the patient is falling away from the normal growth curve.

Regular hearing tests should be carried out due to the high occurrence of chronic middle ear infection (otitis media) and wax build up.

If there is any evidence of heart problems (coarctation of the aorta or heart murmur) a Cardiologist should be consulted.

It is very important that Turner Syndrome girls and their parents be well informed about the condition and its treatment. Support Groups are also very valuable in this respect.

References:

The X's and O's of Turner's Syndrome
Susan Charney — Toronto, Canada
"Good Things Come In Small Packages"
Diane Plumridge, MSW
Crippled Children's Division
The Oregon Health Sciences University

For further information on Turner Syndrome, please contact:

Mrs Glenn Fisher
Turner Syndrome Association Inc
PO Box 112
FRENCHS FOREST NSW 2086
Telephone: (02) 452 4196

ACHONDROPLASIA

There are hundreds of reasons why some children never reach normal height as adults. Many are short in stature because of family or ethnic background. Others have a wide variety of medical conditions, many of them genetic, that seriously limit overall growth, or growth of specific parts of the body, such as the limbs or the torso. Some causes of short stature are well-understood and can be corrected, but most are subjects of ongoing research.

In some cases, individuals with growth defects are extremely short and have normal body proportions. In other cases, they have abnormal body proportions. Among those with abnormal body proportions, some have arms and legs that are very short while the torso is more nearly normal size. *Achondroplasia* is the most common growth defect of this type.

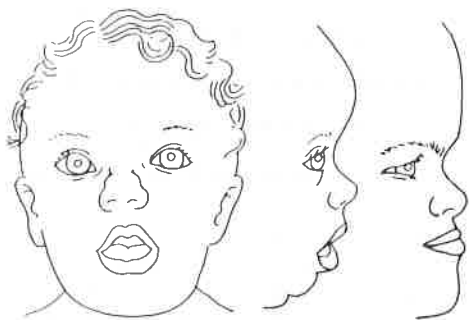
What is Achondroplasia?

Achondroplasia is a genetic disorder of bone growth that is evident at birth. It affects about one in every 10,000 births and it occurs in all races and in both sexes. Its depiction in ancient Egyptian art makes it one of the oldest recorded birth defects.

The word achondroplasia is derived from Greek and means "without cartilage formation", although individuals with achondroplasia *do* have cartilage. Cartilage develops into bone, except in a few places, such as the nose and the ears. Although individuals with achondroplasia have the cartilage growth plates that give rise to new bone during fetal development and childhood, they fail to grow normally.

How Does Achondroplasia Affect A Child?

A child with achondroplasia has a relatively normal torso and short arms and legs. The upper arms and thighs are more shortened than the forearms and lower legs. Generally, the head is large, the forehead is prominent and the nose is flat at the bridge (between the eyes). The large head size is sometimes assumed to be the result of hydrocephalus (excess fluid in the brain), but this is usually not the case. Teeth may be crowded and upper and lower teeth may be poorly aligned.



Depressed nasal bridge,
prominent forehead and lower jaw

A person with achondroplasia usually has a relatively straight upper back with a markedly curved lower spine (lordosis or sway-back). Abnormalities of the vertebrae in the middle back may lead to development of a small hump (kyphosis) in infancy, and to spinal cord compression in adolescence. The lower legs may become bowed, and feet are generally short, broad and flat. Hands are short with stubby fingers that may easily bend backward due to weakened joints. There is a separation between the middle and ring fingers (trident hand).



Short hand with trident fingers

Because of the large head, short arms and legs and loose joints, a baby with achondroplasia is slow to sit, stand and walk alone. This sometimes causes people to think the child is mentally retarded, but most children with achondroplasia have normal intelligence. Other complications include frequent middle ear infections that can cause mild to moderate hearing loss in the first five or six years of life. Low back and leg pains are common, partly because there is pressure on the spinal cord from a small spinal canal. This pressure on the spinal cord can also cause paralysis of the legs, requiring surgery to relieve the pressure.

Children with achondroplasia occasionally have compression of the upper end of the spinal cord, which can interfere with breathing. The compression is caused by abnormalities in the size and structure of the opening in the base of the skull (foramen magnum) and vertebrae in the neck through which the spinal cord descends. Breathing problems also may develop as a result of small chest size, large tonsils and adenoids and small facial structure.

Psychological problems may arise because of the difficulties in adjusting to a world geared to normal-sized people.

What Causes Achondroplasia?

Achondroplasia is caused by an abnormal gene. In some cases a child inherits achondroplasia from a parent who also has the condition. The form of inheritance that governs how the abnormal gene is passed from parent to child is called "autosomal dominant". This means that if one parent has the condition and the other does not, with each pregnancy there is a 50 percent chance that the child will be affected. If both parents have achondroplasia, there is a 50 percent chance, with

each pregnancy, that the child will inherit the condition, a 25 percent chance that the child will not have it, and a 25 percent chance that the child will inherit one abnormal gene from each parent and have severe skeletal abnormalities that are likely to cause early death. A child who does not inherit the gene will be completely free of the condition, and cannot pass it on to his or her own children.

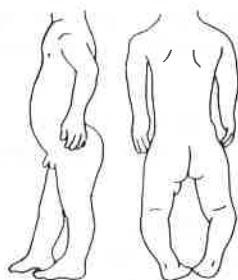
In most cases (over 50 percent) the abnormal gene results from a new mutation (chemical change) that occurred in the egg or sperm cell that formed the embryo. The parents of children with achondroplasia resulting from new mutations are usually average-sized. Typically, they have no other children with achondroplasia, and the chances of their having a second affected child are extremely small. Geneticists have observed that older-than-average fathers are more likely to have children with achondroplasia and certain other autosomal dominant conditions caused by new mutations. Individuals with achondroplasia resulting from new mutations transmit the disorder to their children in an autosomal dominant manner.

Is There a Test For Achondroplasia?

The condition has been diagnosed during pregnancy using ultrasound imaging to monitor fetal leg growth. Most diagnoses are made in pregnancies known to be at risk because one or both parents have achondroplasia, but even in these cases ultrasound examination is not completely reliable. Diagnosis at birth can be made by physical examination.

Can Achondroplasia Be Treated?

There currently is no way to normalize skeletal development of children with achondroplasia. Growth hormone treatments, which increase height in some forms of short stature, do not substantially increase the height of children with achondroplasia, but may increase the disproportion.



Curvature of lower spine,
short arms, short and bowed legs

Infants and children with achondroplasia should be thoroughly evaluated for skeletal abnormalities by a doctor experienced with the disorder. Detection of bone abnormalities that may cause spinal cord compression, leg pain and loss of function, is

particularly important. Kyphosis can be corrected by a small back brace, worn for two years in early childhood. Early surgical correction of leg bone abnormalities can lessen the severity of bowleg deformity. Surgery also may be necessary to relieve nerve or spinal cord pressure from surrounding bones.

Another complication that may need treatment is childhood ear infections. Untreated, these can lead to major hearing loss. Dental problems caused by overcrowding of teeth may require extra routine care and braces, and sometimes removal of one or more teeth.

Can Achondroplasia Be Prevented?

There is no way to prevent the majority of cases of achondroplasia, since these births result from totally unexpected gene mutations in unaffected parents. Genetic counselling can help affected adults make informed decisions about family planning.

What Research Relates To Achondroplasia?

Investigators are exploring mutations of various genes in cells from patients with many of the different forms of short-limbed skeletal maldevelopment, as a basis for devising means of prenatal diagnosis and perhaps eventual attempts at gene therapy. Researchers also have been involved in studying the responsiveness of cartilage cells to hormones important for skeletal growth, and in devising better ways of determining which babies with achondroplasia may develop life-threatening breathing problems.

Are There Additional Sources of Information?

Diagnostic, counselling and management services are provided by the Clinical Genetics Services in each state. In some states there are specific Bone Dysplasia or Connective Tissue Dysplasia Clinics offering special services.

For further information please contact:

The Little Peoples Association of Australia
(LPAA)
The Summit Road
PORT MACQUARIE NSW 2444

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A FAMILY STORY

EARLY INTERVENTION A Parental Experience

Our third child James was born full term but grossly underweight and was termed "dysmature". He also had a talipes (clubfoot). At 5 weeks he was re-admitted to hospital with failure-to-thrive, had hernias repaired and spent 5 weeks undergoing tests and being tube fed as it was found his sucking was inefficient. At 16 weeks of pregnancy no chromosomal abnormalities were reported.

Two years later his chromosomes were re-checked when it was found he had additional chromosomal material, which explained his birth and perinatal condition. There are no other reported cases with his defect. Now at age 5 he is considered to be moderately/severely intellectually disabled. James fell into that group of babies for whom there is no immediate diagnosis at birth but who are obviously at risk because of birth trauma, prematurity, etc. Although medical needs may be superbly met, sometimes these families fall through the net, missing out on available services such as early intervention, welfare worker care, and support groups. In cities these services are available if you know of their existence and you are aware of your need for the service (at the time, not in hindsight!).

I consider it was our good fortune that another mother suggested we visit the Early Education Clinic at Parramatta when he was 9 months, as that association ultimately led to many other services. He was visited by a weekly home-visiting occupational therapist from the Institute for Deaf and Blind Children, North Rocks, and also spent one year in the Department of Education Early Childhood Support Unit. We attended a special playgroup for two years.

The following is a rationale for early intervention gleaned from the two referenced books, and substantiated from personal experience.

Early intervention programs:

1. Enable parents to become more effective teachers of their children.
2. Strengthen the parent's sense of autonomy and self esteem.
3. Respond to the family's needs and thus improve the home environment.
4. Support the parents in the role of caregivers.
5. Facilitate parent to parent information exchange.
6. Help parents to see the child's behaviour from a developmental perspective.
7. Help parents ultimately provide school teachers with information that can contribute to special programming.

Parents are automatically teachers of their non-handicapped children, yet parents frequently need specialised training (invariably uncomplicated) to enable the parent's efforts to be more effective when working/living with a child with special needs. As in the "bonding process", when a parent feels he/she is doing something constructive for their baby, then the acceptance of the handicapping condition may be made easier. In addition when an infant is not stimulated appropriately then the delay becomes manifest in unmotivated children who lack exploratory behaviour, curiosity and play. Not only may they have deficits in social behaviour, but they may develop socially inappropriate behaviour.

In our case we had to realise that severely handicapped children typically learn slowly, maintain newly learned information poorly, and fail to generalise learning adequately.

All aspects of early intervention learning are approached through play in a relaxed and non-threatening atmosphere. Through exposing and teaching an infant a range of fine and gross motor skills, language and personal/social skills he can then employ these skills to occupy himself in meaningful play.

Early intervention has been shown to be cost effective. The cumulative cost of providing special education is less the earlier the intervention begins.

References:

Evaluating Early Intervention Programs for Severely Handicapped Children and Their Families. Ed. Bickman and Weatherford. Pro-Ed 1986, USA.

Working with Parents of Exceptional Children. Ehly, Conoley and Rosenthal. Times Mirror/Mosby College Publishing 1985 USA.

Jan Harrison, Beecroft, NSW

* * * * *

Dear Roslyn,

I am writing you a short letter about Robert and I, for you to publish for anyone to read who is feeling the same things I am about their child.

I'm Not Going Crazy!

Robert was a tiring pregnancy — couldn't wait to go into labour and have him. Well that was a mistake. My third baby was the most awful experience in my life — he changed everything. 14 hours in delivery, face up presentation. Finally he was born. He had problems — only minor, but different to the other children I'd had. He was cold and was taken to the heater for warmth — high pitched screaming all the time.

When I got home it was still the same. Doctor said he is just a bad baby. 4 months old in hospital for FTT screaming and lack of sleep on my part and Robert's. He hasn't stopped since he was born. Just 6 month colic!

Since that diagnosis, Robert has been in hospital 12 times for FTT, enlarged liver and spleen, diarrhoea (chronic), enlarged left kidney, iron deficiency. Still screaming with bad behaviour — has Early Childhood Education at Parramatta.

Robert is 3 years old now and still I don't know anything more about why he is doing this. Doctor thinks I need a Psychiatrist to help me cope with him.

All I want is answers by the doctors. I don't know what to expect anymore. I don't know what is ahead of me and Robert, as no research has been done on Ring Chromosome 21, so they tell me.

Robert and I are trying to get our lives in order and laugh at one another, instead of fighting to get him to drink or eat. It is hard when you are home looking at him not growing, not smiling, not a happy little 3 year old boy he should be. According to the doctor's he is just a normal little boy!

A too caring mother
Name and Address supplied

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If you have a story to tell, please send it into the Editor at your convenience. This is your Newsletter — Health Professionals, your stories also please.

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SEMINARS/WORKSHOPS

The Turner Syndrome Association of New South Wales Incorporated

SEMINAR

29th and 30th September

The Dougherty Centre, 7 Victor Street, Chatswood

GUEST SPEAKERS — SATURDAY

Dr Christopher Cowell — Endocrinologist - Childrens Hospital, Camperdown

Dr Susan Fleming — Gynaecologist and Obstetrician

— Royal Prince Alfred Hospital

Ms Anne Cusick

— Occupational Therapist

— Cumberland College of Health Sciences

— Author of "Choices"

Mrs Ingrid Montgomery — Turner Syndrome Adult and Mother of Taryn

WORKSHOPS — SUNDAY (HALF DAY)

For Turner Syndrome Girls: How I feel about Turner Syndrome

For Turner Syndrome Teens: Growing Up and Relationships

For Turner Syndrome Adults: Accepting your Sexuality

For Parents: Talking to your Daughter about T.S.

Open: Practical Advice on Growth Hormone

Open: Fashion and Makeup

Open: "Food Glorious Food" and Healthy Eating

Enquiries: Mrs Glenn Fisher, Telephone: (02) 452 4196

PO Box 112, Frenchs Forest 2086

MAKE A NOTE IN YOUR DIARY NOW! September, 18th to 21st, 1990

National Council on Intellectual Disability

will hold a

NATIONAL FAMILY FORUM

"Answering the Dilemma of Change"

at the Country Comfort Inn, Canberra

This Forum will bring together

- families • service people • advocacy groups •
- general community organisations • relevant government officials •

to find better solutions for the problems that face families at this time of uncertainty and change

The structure of the Forum is being determined by parent's concerns. If you would like your concern to be addressed at the Forum, let us know.

During the conference a half-day workshop will be conducted by Brian Salisbury of the G Allan Roeher Institute, Canada on: *Service Brokerage and Individualised Planning*

All enquiries about the Forum should be directed to:

National Secretariat, GPO Box 647, Canberra ACT 2601

Phone: (06) 247 6022

CONTACT CORNER

AGSA will publish requests for contact and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The Editor may be contacted for the information to be passed on in the case of anonymous requests.

Privacy and anonymity will be ensured if requested.

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While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgemental approach is recommended in establishing contact.

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Request from Education Seminar — Mother of 6 year old girl with ORAL FACIAL DIGITAL SYNDROME (OFD) Type 1 seeking contact.

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Cri-du-Chat — Request for other families similarly affected.

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Request from the North Coast — Community Nurse seeking contact for a family with Tay-Sachs.

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Contact A Family — UK looking for contact with Bloom's Syndrome. Can anyone help with this request?

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Mother seeking information and contact for 20 year old son suffering from COFFIN-LOWRY SYNDROME.

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Two ladies — one on the South Coast (aged 34 years) — one in Sydney seeking contact with others and their families with SPINOCEREBELLAR DEGENERATION. This disorder causes imbalance/mobility problems thus the sufferers have difficulty with writing and walking. Anyone willing to 'phone or write?

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Is there a contact or information for a family with a nine year old girl suffering from SHPRINTZEN'S SYNDROME?

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Can anyone help with a contact for LEIGH'S DISEASE? Mother would dearly love contact and information.

FROM CONTACT A FAMILY — LONDON

In response to the enquiries featured in your contact corner for the November 1989 issue of AGSA Newsletter, I wonder if the following are of help.

1. Ring chromosome 21, Deletion of chromosome 15 or Monosomy 15

Mrs Edna Knight
Rare Chromosome Support Group
160 Locket Road
Harrow Weald
Middx HA3 7NZ
ENGLAND
Telephone: 081 863 3557

2. Fraser Syndrome

Mr and Mrs Cooper
20 Hillbrow
Bearsted
Kent
ME14 4AN
ENGLAND

The child of this family has died but Mr and Mrs Cooper happy to have contact.

3. Non-ketotic Hyperglycinaemia

Research Trust for Metabolic Diseases in Children
53 Beam Street
Nantwich
Cheshire CW5 5NF
Telephone: 0270 629782

4. Dejerine - Sottas Syndrome

Jenny Lee
Department of Social Work
The John Radcliffe Hospital
Off Headley Way
Headington
Oxon OX3 9DU
ENGLAND

5. Goldenhar Syndrome Support Group

Aileen Wilson
17 Ardgowan Street
Greenock
Renfrewshire PA16 8LG
SCOTLAND

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Closer to home — contact has been established between the family of "No 2 chromosome defect specifically in vicinity of 2q21 to 2a32", and a family in Queensland with an abnormality in the No 2 chromosome.

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QUESTION AND ANSWER COLUMN

Some interest has been shown in a Question and Answer Column for the Newsletter, eg

Q. What is trisomy 21?

A. Trisomy 21 is, in fact, another term for Down Syndrome.

All answers will be thoroughly researched by our Medical Liaison Committee.

If you have a question(s) please send them to the Editor, 44 Rawson Street, Epping 2121. Your request will be published where and when space allows.

Please ensure that you send your name, address and telephone number (clearly written or typed) thus enabling any queries to be dealt with speedily before printing in the Newsletter.

SUPPORT GROUP

NEWS BULLETIN

Your chance to publicise and provide information about your Society's or organisation's upcoming events and new developments.

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The following is a list (of **AGSA Member Support Groups**) for your information:

Spina Bifida Association of NSW
 Cornelia De Lange Syndrome Support Group
 The Little People's Association of Australia
 Fragile (X) Support Group
 The Williams Syndrome Association
 The Australian Tuberous Sclerosis Society
 The OI Society of NSW
 The Neurofibromatosis Assoc. of Australia
 Spinal Muscular Atrophy Support Group
 The Dystrophic Epidermolysis Bullosa Research Association
 The Society for Mucopolysaccharide Disease (MPS)
 The Turner's Syndrome Assoc. of NSW
 Retinitis Pigmentosa Society of NSW
 Coeliac Society
 Australian Huntington's Disease Assoc. (NSW)
 Australian Huntington's Disease Assoc. (QLD) Inc.
 Australian Marfan Syndrome Association
 Cystic Fibrosis Association of NSW
 Muscular Dystrophy Association of NSW
 Haemophilia Society of NSW
 ALS — Motor Neurone Disease Association Inc.
 Friedreich's Ataxia
 Handital (Association for Italian Families and Friends of Handicapped Children)
 Down Syndrome of NSW Inc.
 Ehler's-Danlos Syndrome Association
 Lowe's Syndrome Association Inc.
 Thalassaemia Society of NSW
 Albino Support Group
 Spina Bifida Association of WA (Inc.)
 Wellington Huntingtons Disease Association (Inc.)
 Prader-Willi Syndrome Association
 Charcot-Marie-Tooth Disease/Peroneal Muscular Atrophy International - Canada

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Victorian Marfans Syndrome Support Group

CONTACTS

Joy Redstone, Liaison Officer
 PO Box 221
 IVANHOE VIC 3079
 Bruno Caruso, Committee Member
 5 Winifred Street
 PASCOE VALE SOUTH VIC 3044

COST

\$15.00 family membership/donation

ACTIVITIES

Mutual support - monthly meetings
 Community Education - leaflets to medical centres etc
 Information Exchange - quarterly newsletter
 Lobby - for funding
 Individual Advice - resource information
 Services - counselling through the VICTORIAN CLINICAL GENETICS SERVICES

Research - liaising with the Murdoch Institute within the Royal Childrens Hospital

As a Victorian Support, we would also welcome any enquiries from interstate, if a Marfan Support Group structure is lacking in your own area.

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New Address Albino Support Group

PO Box 123
 HABERFIELD NSW 2045
 Telephone: (02) 799 6972 — Averil Legg

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Contact for Rett Syndrome Association of Australia Inc

Mr Bill Callaghan, President
 7 Charles Street
 BELMONT VIC 3216

* * * * *

Charcot-Marie-Tooth Disease (CMT) at present in the process of establishing a group here in Australia.

Contact Person

Ms Elizabeth McDonald
 1 Gibbs Street
 CROYDON NSW 2132
 Telephone: (02) 747 6268 (Home)
 (02) 646 6852 (Work - Tue, Wed, Thu)

* * * * *

similarly Russell-Silver Syndrome

C/- PO Box 153
 COONAMBLE NSW 2829

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Batten's Disease Support Group

(Mrs) Helen Vickers
 12 Nannine Place
 FISHER ACT 2611
 Telephone: (06) 288 0078

Helen has written an excellent Booklet on Batten's Disease which may be obtained by writing to the above address.

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To those Support Groups sending in Newsletters — thank you. They are great reading and it is hoped that you wont be too surprised to actually find an article from your Newsletter appearing in AGSA.

Congratulations to the Friedreich's Ataxia Support Group ... their Newsletter has a name

F A N (Friedreich's Ataxia Newsletter)

FAN — TASTIC! FAN — TASTIC!

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Please send in articles you feel would be appropriate for the Newsletter (typed is wonderful but not essential) — personal stories (people like to know how others are coping or not coping), poems, queries, research updates (professionals please note), state genetic service activities, D.D.S. activities, Support Group Conference dates — in fact, anything you think could be of interest to our readers. Putting together a Newsletter of this magnitude requires the co-operation of all — professionals and lay people alike.

Finding articles is not always easy and I would be most appreciative of your help.

If you have knowledge, let others light their candles at it.

— Margaret Fuller

RESOURCES & Aids

The SPECIAL EMPLOYMENT GROUPS Section of the Department of Employment, Education and Training (DEET)

Our Role:

Advocate for the employment, education and training needs of people with disabilities in the development and implementation of DEET programs and services.

Provide information to CES specialist staff about disabilities and their implications for employment, education and training, and about organisations that support, represent, advocate for or provide services to people with disabilities.

Programs and Services of the Department of Employment, Education and Training (DEET)

For further information contact your local CES office. The CES has special Placement Officers and Counsellors who can give information, advice and referral.

JobStart

This wage subsidy program provides eligible employers with a part re-imbursement of wages for the part-time or full-time permanent employment of persons with disability. It is paid for a negotiable period up to 20 weeks, and rates reflect the relative disadvantage faced by the jobseeker.

Disabled Apprenticeship Wage Subsidy Scheme

Another wage subsidy program which provides an eligible employer with a partial re-imbursement of the wages of an indentured apprentice with a disability. It is paid for the entire duration of the apprenticeship.

Modifications of Facilities

Eligible employers may receive a grant of up to \$2,000 (currently under review) for additional costs incurred in leasing, purchasing or modifying essential equipment and/or modifying the workplace to enable a person with a disability to undertake employment. This program is a component of other DEET wage subsidy programs.

SkillShare

Under the SkillShare program, community-based organisations are funded to provide training. The aim is to assist long-term unemployed and other disadvantaged people to obtain employment or to proceed to further education and training.

There are currently 139 projects across New South Wales and the Australian Capital Territory, offering training in a broad range of vocational skills. SkillShare projects are encouraged to meet the training needs of unemployed people with disabilities.

JobTrain

This program funds industry-based training in a broad range of occupations. Courses are provided by TAFE and private sector trainers. Many people with disabilities have attended JobTrain courses and have the skills required to get jobs.

JobSearch Training Courses

These are short courses for young people which concentrate on job getting skills.

JobClubs

JobClubs provide intensive assistance in job getting.

Small groups of job seekers, work together with the support of a co-ordinator and each other, and resources such as telephones, newspapers and word processors.

Australian Traineeship System

Traineeships entail twelve months of work experience and off-the-job training, for people 15 to 18 years of age (15 to 19 for people with a disability) in non-trade occupations in industries such as: Retail, Motor Vehicle, Textile, Clothing, Printing, Insurance, Legal, Plastics and Timber. The range of occupations is increasing.

When people with a disability are employed under this system the employer will receive a subsidy to offset the cost of on-the-job training; financial assistance for the purchase of special equipment or necessary modifications to the workplace environment; and reimbursement of the cost of tutorial assistance if a trainee with a disability requires additional training.

Trainees are entitled to up to one hundred hours of additional tuition to assist with off-the-job training. Blind or deaf trainees are entitled to tutorial assistance for the length of the off-the-job training and to interpreter assistance if required.

Income Support Programs

Austudy — Income support for eligible students who are studying full time at secondary school or a tertiary institution.

Formal Training Allowance — Income-tested support for eligible trainees doing approved DEET training courses.

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ASK CID Information Service

NSW Council for Intellectual Disability



Information Service

The NSW Council for Intellectual Disability has developed a substantial information base called ASK CID — about disability services in New South Wales.

ASK CID will help people with an intellectual disability, their families, and their carers, to keep in touch with everything that is available to them throughout the State.

The data includes accommodation, employment, leisure activities, specialised travel agents, developmental disability teams, community centres, ethnic contact points, holiday programs, respite care, parent support groups, activity therapy centres, early intervention centres, regional education officers for people with an intellectual disability, TAFE officers, and much more.

How can this help you?

We hope you will ring or write for any information you need. For example, you may want to know about respite care at Lismore; employment options in the Parramatta area; you may be planning a special holiday.

We will, most probably, be able to help you straight away! The data is stored on computer to make it readily accessible. New data will constantly be added, and old information will be regularly updated.

We are happy to provide information to anyone seeking it — with priority to members.

For further information contact:

Alison Price
NSW Council for Intellectual Disability
4 Doig Avenue
DENISTONE EAST NSW 2112
Telephone: (02) 807 1411
Facsimile: (02) 809 7135

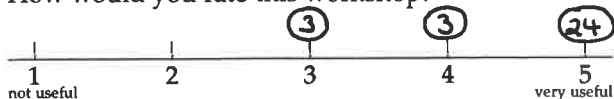
SEMINAR EVALUATION

The "Education — and you" Seminar held on 2nd June at Epping RSL was attended by approximately 80 people.

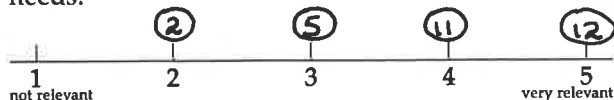
The following is an extract from the Evaluation Forms (not completed by all) which may be of interest to those who were unable to attend.

Evaluation Form

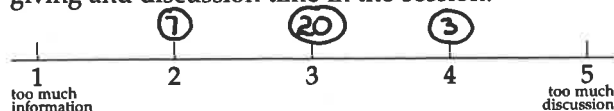
1. How would you rate this workshop?



2. Was the information presented relevant to your needs?



3. Was there a good balance between information-giving and discussion time in the session?



4. What were the main benefits you got from this workshop?

- Differences between three education systems and overview given (2)
- Hearing other parents' experiences
- Meeting a lot of good people
- Confirmed my gut feeling that the Department of Education is not adequately providing a service in special education
- Short talk on siblings
- Point made as to the importance of contacting integration officer!
- The information other parents had about schools for their child
- Understanding what is available in education for disabled children (9)
- We now know where to start in schooling. We have phone numbers and knowledge where to go for help. I'm not so frightened and flustered about having to face this on my own.
- We got some very helpful information (3)
- Being informed of information I didn't know re Department of Education which would have been helpful at the time of my son's placement. The hope that things are on the improve though obviously not quickly enough.
- The importance of Parent Support Group/Advocacy group
- Sharing of difficulties, emotions and fears (3)
- Reinforcing my knowledge (2) and decisions about education
- Names/position of relevant contact within education systems (2)
- Starting point for seeking further information
- Deciding what type of education my child needs (3)
- Communication with others
- Reinforcement that the scholastic need of the child was more important than the social skills previously suggested.
- Put education first — then think of other issues
- Be more assertive in decision making
- Made me decide to re-evaluate the direction for my 11 year old's education **now** instead of waiting for something to go wrong in the future

- Points I can raise with the teacher of my son in a support class, so as to improve integration with other children
- The opportunity to speak to Mr David Hilder and thus ensure the course of action being taken with my children is correct
- To make a decision early and to contact regional officer

5. Were there any improvements that could be made to the workshop?

- Beginning with a speaker on early intervention would have been beneficial
- Grouping people with children with similar disabilities to discuss education options. I feel all levels should be integrated but some people don't!
- An excellent day
- More time given to the visiting speakers would give more time for specifics and questions (8)
- More discussion time (5)
- Perhaps people could be told to write down their questions and follow up later outside of workshop
- More variety of discussion group questions so we can share different ideas at feedback times
- Workshop was too restrictive — would have preferred a more personal discussion
- Perhaps the workshop tried to cover too many topics/issues in available time — is it possible to provide seminars which are more specific?
- A little slow to start
- I don't feel we needed notes written up during discussion summary — it slowed things down
- Address the subject of voluntary care and how to obtain it
- Summary sheets from various education areas, ie as on overheads presented
- Perhaps have as speakers, parents of children that have been integrated and those in special schools
- Further information for physical disabilities
- I would have liked the discussion group early in the day — as I came by myself, meeting and talking to other people was very good
- Advice for people who have pre-school children — what is available in resources

6. Are there any other comments you would like to make?

- I think people in the country areas need more support because for 5 years until I moved to Sydney I had no support. Maybe hospitals need to be told about groups.
- Would have really liked to discuss with Education Department spokesman about "least restrictive environment". Not happy with the policy — "provided there is enough support for the child" — and then not enough funding for Special Education Teacher support-policy is very subjective and covers the department well.
- A well-balanced day. I enjoyed it.
- I believe after today a lot of people, including my husband and I, have had a load taken off their shoulders. Putting our child in school frightened us. We didn't know where to start, now we have options. I would also like to know how I can air my beliefs outside my four walls, say to the media etc ...
- (Workshop was) a very valuable resource for myself and parent support group.
- Documentation available very beneficial
- A chance to mix with parents from other areas and exchange ideas

- Hope more of these days are held on varying subjects
 - I would like to know what is available for a blind child if she does not get into North Rocks
 - Maybe some "Special Needs" children of different ages could speak to small groups on how they feel about their school
 - Include information on early intervention — it is very important that parents realise they can help their child learn before school age
 - Thanks for the opportunity to attend — hope you are successful in reaching all people who require your services
 - I feel the panelists were too rushed in delivering their talks — it seemed they had more to say and could have taken more questions
 - There is a need for more information for hearing impaired children (2), also "mild" people (1)
 - It was a very interesting and informative day (4)
 - I would like the chance to meet with a group of Department of Education people — not just one — and to talk in depth regarding the real difficulties that parents with disabled children are coming up against in finding a placement and with transition and on-going support
 - I am glad I came, it is a pity people either didn't know about the day or didn't come
 - I particularly enjoyed hearing Lee Mills talk. I also had an opportunity to talk to David Hilder about two problems I had last year with the Education Department and it was valuable to get his answers as I will act on that information this week.
It was great to get a copy of the Special Education policy and disability enrolment procedures
 - It's beneficial to meet with other parents who have the same interests and concerns
 - Thank you for a professionally run seminar (3)
 - Thank you for the information you gave us — we now have something to go on with when checking schools for our son
 - I got more out of today than I have in the past six years
 - Location/cost/timing of this workshop was very good — don't change!
7. Do you have any suggestions for future workshops?
- Early intervention
 - Lobbying for improvements in Education system
 - Effective leadership in support groups
 - Effects on siblings and family
 - Small groups discussing each other's children meeting and talking to people with similar handicaps — helping one another from our experiences
 - Independent living for the disabled child (3)
 - More information for severely disabled children
 - Ways parents can cope with stress of handicapped children: What comes next? After school.
 - What is available at Secondary level of education? — Secondary education in general (perhaps TAFE) for disabled
 - After school — What? (ie after 18)
 - How/when do you explain to your child/siblings that they have/their sibling has a disability
 - Coping with child's school problems. How do you help the child to cope?
 - Maybe a "mens only" workshop for the fathers on the problems they encounter having a disabled child in the family
 - More about hearing impairments

- How to cope with family denial, ie when entire families disown mother/father and disabled child
- Discussion on advantages and disadvantages of mainstreaming for mild disabilities
- Repeat this each year as we'll need to be informed about changes to each of the 3 options of education and new parents will always be looking for this information
- What to expect in a high school with an integrated support class — issues to raise with principal etc
- Integration
- Setting up Parent Support Group
- Assertiveness training
- Workshops should be held more often with up to date information
- Seminar for pre-schools. What's available for your child who needs intervention?

If anyone requires further information re this Seminar or subsequent Seminars, please contact

Mrs Margaret Cupitt
Co-ordinator
The Family Education Unit
Telephone: (02) 805 0233

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RESPITE CARE

Riverlink InterChange Inc
PO Box 281
EASTWOOD 2122
Telephone: (02) 858 2233

covers the Lower North Shore and Ryde-Hunters Hill Respite Care.

What is Respite Care?

The term "Respite Care" means relief care or "time off". It is the provision of care to a child or young person with a disability. Respite Care provides a period of relief to the primary caregiver and family while broadening the child or young person's social network and encouraging independence.

What does "Disability" mean?

A disability can be either physical, intellectual, sensory or emotional and may impair the individual's development and learning.

Riverlink provides Respite Care for children and young people with disabilities ranging from mild to severe.

How is Respite Provided?

Respite Care is provided by host families within the local area. The child or young person is cared for in the host family's own home.

Why is Respite Care Needed?

The purpose of the Service is to meet the varying individual needs of children, young people and their families:

i) For Parents and Family

Respite Care allows parents and family the "time off" from the constant demands and supervision involved in caring for a child with a disability. These families may not receive the same help and support as do families of non-disabled children. People feel that caring for a child with a disability is different from that of a non-disabled child and that they would be unable to cope. Consequently, they may not offer their support. Respite Care provides the much needed time off which every family needs.

ii) For the Child or Young Person

Often a disability excludes the individual from the same opportunities as a non-disabled child to form a social network outside their own family, to gain new social experiences and develop a sense of independence. Riverlink Interchange helps to meet these needs.

What Does Being A Host Family Involve?

A "host family" can be a traditional two parent family, single parent with or without children — any person who is interested in caring for a child. Orientation and continual support is provided by Riverlink to host families. It is important to remember that host families are part of a team.

For host families, involvement in the Riverlink programme is an opportunity to make a new friend and become an important part of their life.

What Types Of Care Are Required?

The period of care is limited short term care usually between 2 hours to 2 weeks. The length of time and frequency of care is negotiated between the natural family and host family. What the care of the child will involve depends upon the individual needs of the child. To ensure that the child's needs are met, the child is carefully and appropriately linked to a host family.

FAMILY BASED RESPITE CARE VICTORIA

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Ms Jane Stickels — Co-ordinator
Ms Trix Vandeheuvel — Field Worker
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CANTERBURY 3126 (03) 836 9811

Ms Myrta Iten — Co-ordinator
256 Murray Road
PRESTON 3072 (03) 478 9633

Ms Vera Rudinica — Co-ordinator
9E Anderson Street
PO Box 538
PASCOE VALE SOUTH 3044 (03) 350 4600

Ms Lyn Soltani — Co-ordinator
Suite 18A, Bayswater Shopping Village
BAYSWATER 3153 (03) 729 5422

Ms Maggie Matheson — Co-ordinator
Ms Hazel Wilson — Co-ordinator
265 Bluff Road
SANDRINGHAM 3191 (03) 598 1963

Ms Marci Baron — Co-ordinator
Ms Diedre Tellefson — Field Worker
69 Victoria Street
FOOTSCRAY 3011 (03) 689 6644

Mrs Jenny George — Co-ordinator
Mrs Dianne Wright — Field Worker (Asst Co-ord)
Ms Karen Lott — Field Worker
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PO Box 672
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Mrs Carole Benham — Co-ordinator
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MORWELL 3840 (051) 34 6225

Ms Irene Kloessmann — Co-ordinator
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DANDENONG 3175 791 7066

Ms Christine Volk — Co-ordinator
30 Raglan Street
PO Box 145E
NORTH BALLARAT 3350 (053) 31 2115

Ms Judy Rasmussen — Co-ordinator
Mrs Linda Penny — Field Worker
Kilmany Family Care
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Mr Des Moyle — Co-ordinator
Ms Hilary Guest — Field Worker
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BENDIGO 3500 (054) 42 1766

Ms Rhonie McIvor — Co-ordinator
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QUEENSLAND

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Sunshine Coast Family Link
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NAMBOUR 4560 (071) 41 6600

Debbie Cumming
Westcare
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PADDINGTON 4064 (07) 368 1272

Ane Marie Byrne
Host Family Scheme Co-ordinator
270 Roma Street
BRISBANE 4000 (07) 229 5888

Rebecca Storey and Nerida Boyle
Mamr'e Family Link
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CLOVELLY PARK 5042 (08) 277 8566

Hans Peters
Home and Community Care
ADELAIDE (08) 213 3499

NORTHERN TERRITORY TASMANIA

AUSTRALIAN CAPITAL TERRITORY
NEW SOUTH WALES METROLOLITAN/COUNTRY
NEXT ISSUE

AGSA aims to:

- educate the medical and allied health professionals and the community about genetic disorders
- lobby government bodies, both Federal and State, for appropriate funding for genetic services
- provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group
- facilitate accessibility to individual support groups for those families with a particular genetic disorder
- provide a forum for the exchange of information between support groups regarding available community services.

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