THE · ASSOCIATION
OF·GENETIC·SUPPORT
OF · AUSTRALASIA

FUNDED BY THE NEW WALES HEALTH DEPARTMENT

OCTOBER 1995 ISSUE 20

MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

EDITORIAL

It is hard to believe that another year has flown past and we have now embarked upon a new financial year for AGSA. A current year (95/96) Membership Form is included with this Newsletter.

As indicated in an earlier Newsletter, a number of Management Committee positions will become vacant at our Annual General Meeting to be held in early 1996 and we would encourage you to consider nominating for one of these positions. Remember, only financial members are eligible to vote.

Should you have any queries regarding the role of Management Committee please feel free to contact me on (047) 515 872 or Dianne at the office on (02) 211 1462.

Wagga Wagga Seminar

For number of years, outreach to country areas has been very much the concern of the AGSA Committee. Recently we held a most successful one day Seminar in Wagga, entitled "Rural Genetics Support Seminar" organised by our Regional Representative, Judy Rands. This Seminar was attended by both professionals and family members and covered a diverse range of topics from prenatal diagnosis - what options are there/what does it all mean - to assertiveness, coping skills and strategies.

Evaluation of the day's activities indicated an on-going need for this type of seminar and of particular significance was the enormous amount of networking that went on between participants. However, support and information were still the highest priority for families (see Regional Representative's report).

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Thank You
On a more personal level I would like to thank all those AGSA members who wrote rang or sent flowers following the death of my father on 19th August. Your expressions of sympathy are deeply appreciated.

ROS SMITH

PEER SUPPORT/INFORMATION OFFICER'S REPORT

The Rural Genetics Support Seminar at Wagga Wagga was very successful and I enjoyed making contact with the families and health professionals who attended and it was good to meet Bernadette Taylor who is the genetic counsellor for area. As a consequence of the meeting two ladies in their fifties both with the same and very rare condition have now been put in contact with each other. Coincidently they live only two hours apart and both thought they would never meet another person with the same condition (see Regional Representative's Report).

On Sunday 29th October approximately 20 people attended the first **Lysosomal Storage Disorders** meeting. The speakers were Prof. John Hopwood, Dr. Meredith Wilson, Mrs Ros Matthews (Gaucher parent), Mr David Lewis (Genzyme) and Mrs Laura Greffe (Gaucher Association of Australia). Following a round table discussion the group unanimously agreed to the formation of an umbrella support group.

On Sunday 12th November there will be a follow-up **Haemochromatosis** meeting to be held at the AGSA office commencing at 10 am.

The profile this month is on **Velo Facial Cardio syndrome** and on Friday 3rd November 1995 Dr Alison Colley, a Clinical Geneticist from Hunter Genetics spoke on "VCF and Di George syndrome. Are they the same?" at Hillview Community Health & Resource Centre, Turramurra.

Recently AGSA has had a number of people drop in at the office requesting information. Hopefully, in the New Year we will be setting up a small reference library. Also AGSA hopes to provide support to the smaller groups by way of use of a computer and a photocopying machine. This will depend on AGSA obtaining extra office space in the building. AGSA would like to thank Ross Rayne and Rodney Nash of Canon Australia Pty Ltd and Gene Parmenter of Muscular Dystrophy for the kind donation of a Canon photocopier. AGSA looks forward to sharing this donation with our members.

Please contact me with any queries.

Best wishes

DIANNE PETRIE

REGIONAL REPRESENTATIVE REPORT

On Saturday 16th September, 1995, the first **Rural Genetics Support Seminar** was conducted at Wagga Wagga. This seminar was conducted over a full day and covered topics on Genetic Counselling, Prenatal Diagnosis, Assertiveness Skills, Coping Strategies, Long Term Care and Respite Care, AGSA's Role and the Role of Support Groups.

There were twenty participants and from general comments on the evaluation form, and during and after the seminar, the day was well received. Some of the participants travelled some distance to attend the seminar with people coming from Young, Wodonga, The Rock, Junee, Albury, Lavington, Wahgunyah, Yerong Creek, Lockhart and of course, Wagga Wagga. There were a mix of parents of children with genetic disorders, persons who themselves have a genetic condition and professionals. There was ample time for networking and the participants made good use of this time.

The speakers covered their topics well and allowed for group participation. They were available for discussion following on from their session, with some of the speakers remaining for the entire day.

The last session of the day was a round table discussion and included future planning for this region. There was a request for a similar seminar to be conducted in Albury perhaps next year. Further discussion and evaluation will occur regarding this matter.

Overall the seminar went very well and was enjoyed by all. I would like to thank all those who helped prior to, and on the day of the seminar. Planning will begin again in the New Year for the next regional seminar, any ideas please contact me.

Judy Rands,

Regional Contact,

(069) 261-560.

MEETINGS

TAAG PICNIC/ANNUAL GENERAL MEETING SUNDAY 19TH NOVEMBER 1995.

Pecky's Playground,

Reservoir Road

Prospect NSW

Time : NOON

BBQ facilities available.

For further information contact Marie Taylor (02) 9938 4726

FAMILY ADVOCACY INVITES PARENTS AND FAMILIES OF TEENAGERS AND ADULTS WITH DEVELOPMENTAL DISABILITIES TO:

"INCLUSIVE LIVES"

Venue: City of Sydney RSL Club,
565 George St, Sydney.

Date: Thursday 23rd November 1995

Time: 9 am - 4 pm (PH: 869 0866)

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CONTACT CORNER

AGSA will publish requests for contacts 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 AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

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-judgmental approach is recommended in establishing contact.

APERT SYNDROME

AGSA is seeking contacts for two families who have children with this condition. Please contact AGSA`s office if you know of other families.

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CHROMOSOME 11 DELETION

(11q 23q ter)

A mother of a seven year old girl would like contact with a family with a similar condition. Please contact AGSA for details.

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DOWN SYNDROME/AUTISM

A mother of a nine year old boy would like contact with a similar family. Please contact AGSA for details.

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HALLERMAN-STREIFF SYNDROME

Parents of a two month old baby boy would like contact. Please contact AGSA for details.

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HEREDITARY ANGIONEUROTIC EDEMA (HANE)

A mother who has two boys with this condition would like to meet up with others for support. Please contact AGSA for details.

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MITOCHRONDIAL DISORDER

A mother of a 14 month old baby girl would like contact with another family. There used to be a support group but it is no longer functioning. If anyone knows about this group or a family please contact AGSA.

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REPENNING SYNDROME

A Queensland family with two boys with this condition would like contact with another family. Please contact AGSA for details.

WILSON'S DISEASE

A lady with this condition would like to meet up with another person. Please contact AGSA for details.

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SUPPORT GROUPS

THE AUSTRALIAN CHAPTER OF THE BATTENS DISEASE SUPPORT AND RESEARCH ASSOCIATION

For Information Please Contact:

Harry & Lynne Partridge

32 Coronga Crescent

KILLARA NSW 2071

TEL/FAX (02) 418-2043

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(The following letter is an excerpt from the St George & Sutherland Shire Leader, Thursday 26th October 1995, reprinted here with permission from the writer)

CALL TO HYDROCEPHALUS VICTIMS

As a person with hydrocephalus, I agree with your recent article on the lack of funding and research into this condition.

There is no association in Australia for people with hydrocephalus, who are at present referred to the spina bifida group or association nearest to them, whether they have spina bifida or not.

In August 1992, I set up an informal group known as HEAD, or Hydrocephalus Education, Action and Development.

We now have about 40 full members and several family members.

Our age range is from 12 months to 64 years. We also take people with spina bifida, as long as they also have hydrocephalus. Our first meeting will be held soon.

From the article, I read that there are about 45 people in St George and Sutherland Shire with this condition. I would be pleased to hear from them, or anyone else who is interested.

Penelope Wilkinson

13 Hosking Crescent, Glenfield 2167

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THYROID FOUNDATION OF AUSTRALIA

A year ago Dr Steven Boyages, Clinical Director of Department of Diabetes and Endocrinology at Westmead Hospital, met with a small team of interested people regarding the start of a **Thyroid Support Group.**

Abnormalities of the thyroid gland are common and affect about 1 in 40 of the population. This may be as much as 1 in 20 or 25 of the general population if small thyroid nodules are included. Thyroid disorders are much more common in women than in men and some thyroid disorders have a familial tendency. Autoimmune disorders of the thyroid are common. These are Graves' disease and Hashimoto's thyroiditis: each effects about 1 in 100 of the population. Other less common causes of thyroid disease include nodules, thyroid cancer, thyroid inflammation and primary hypothyroidism (under activity).

There are several thyroid support groups overseas which produce information materials for patients which have been extremely useful for people with thyroid disorders in Australia. Following the launch of the Australian Pituitary Foundation in March 1994 it was felt that it was time that disorders of the thyroid, in particular, required special attention with a support group focus.

In April 1995, Dr Steven Boyages had an article entitled "Tired, depressed, check your thyroid" published in the Readers Digest. This article generated an enormous amount of enthusiasm and over 200 people contacted the department for further information regarding a support group.

On 29th October 1995, the Department held a thyroid seminar at Westmead Hospital as an introduction to launching the Thyroid Support Group. The seminar had the format of an interactive workshop about the thyroid gland and participants were encouraged to take part by asking questions and becoming involved on all aspects of thyroid disease. Speakers at the seminar were Lyn Taylor, Senior Nurse Manager of Diabetes and Endocrinology Ambulatory Care Centre and Dr Steven Boyages, Director of Diabetes and Endocrinology Department. An overview of the thyroid gland was given and its relationship to the pituitary gland. Other areas covered were hyperthyroidism (over activity). hypothyroidism (under activity), thyroid nodules and cancers.

Members of the thyroid support group gave an overview of their aims which are to:

* educate to raise the awareness of thyroid disease
* support patients and families
* lobby regarding funding for thyroid disorders and further research and
* increase membership of the Thyroid Foundation.

One hundred and four people attended the seminar from all areas of NSW, Queensland, Canberra and Melbourne. There was enormous enthusiasm from the audience regarding interest in thyroid disorders and a willingness to participate in a thyroid support group which gave the opportunity of reducing the isolation factor of having what is usually a chronic condition.

Genetic predisposition to thyroid disorders is very rare however there is a strong familial element with some thyroid conditions which raises the need for advice and reassurance as particularly important.

There will be further thyroid seminars held at Westmead and a continued effort to increase membership and inform the public of this new support group.

It is planned that the NSW Health Calendar for 1996 will list "Thyroid Awareness Day" for 26th October.

GENETIC RESOURCE LINE

Washington State has taken the lead in outreach to primary care professionals by establishing a toll-free Genetic Resource Line. The GRL, a joint project of the University of Washington and the Washington State Department of Health, will not only provide information about issues in medical genetics such as diagnostic testing, genetic counselling, support groups, and cultural and ethical issues, but will also be a resource for information about regional continuing education opportunities. All calls are answered by a specially trained staff. It is not designed to answer consumer calls. The number is 1/800/562-GENE.

CLASSES

Stage Coaches

Dance is becoming better recognised as a form of physical, social and personal therapy for children and adults with disabilities. **Majella Barbe** is a paediatric occupational therapist based in Double Bay with dance teaching experience Brisbane, Sydney and the UK. Majella hopes to establish local groups for children with mild to moderate intellectual, physical, sensory and learning disabilities, and would like to hear from local parents who may be interested in group or individual sessions (326 1493 after hours).

Matthew Beer is an established drama teacher experienced in working with people with disabilities. Matthew has a timeslot available in his packed schedule for a new class from 10 am-12 noon every Tuesday at the Pyrmont Community Centre, cnr John & Mount Sts, Pyrmont. Drawing on his experience in teaching drama to adolescents and adults with developmental disabilities, Matthew would like to hear from anyone interested - he intends to shape the class to suit participants' interests, needs and desires (692 9178).

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CONFERENCES

Genetics in the Classroom: A Conference for educating professionals about genetic causes of developmental disabilities - Nov 17; Philadelphia, PA. Presented by Elwyn, Inc., and the Mid-Atlantic Regional Human Genetics Network. Contact Elwyn, 111 Elwyn Road, Elwyn, PA 19063.

Hemifacial Microsomia Symposium

April 22-23 1996; San Diego Princess, San Diego, CA. Contact American Cleft Palate-Craniofacial Association, 1218 Grandview Avenue, Pittsburg, PA 15211; TEL 412/481-1376

PROFILE A-Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes 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.....

VELO-CARDIO-FACIAL SYNDROME

This information was kindly supplied by Dr. Tony Lipson and the VCFSFA.

Velo-cardio-facial syndrome (VCFS) was first described by Dr. Robert Shprintzen at the Montefiore Hospital and Medical Centre, in New York in 1978. This is why it is sometimes called Shprintzen syndrome. "Syndrome" means a pattern of features occurring together.

In the last fifteen years there has been significant research on VCFS and the identification and treatment of children with the syndrome. This pattern is also known as Di George, Cardiofacial syndrome and Conotruncal Anomaly Unusual Face syndrome. It is an extraordinarily variable pattern with no single feature being exclusive.

VCFS is a genetic condition associated with a deletion of a small segment (usually submicroscopic) of the long arm of one chromosome 22. As a result of the deletion, some genes are absent from the chromosome. Humans have 22 pairs of chromosomes, numbered 1 to 22 and two sex chromosomes making a total of 46. The 22nd chromosome is the smallest chromosome. The deletion on chromosome 22 is present from conception.

In most cases the parents of a VCFS child do not themselves have VCFS and there is no known reason for the deletion occurring in the child's number 22 chromosome. However in 10-15% of cases it is inherited from a parent with VCFS. When a parent has VCFS the chance of their child having VCFS is one in two, or 50/50 in every pregnancy.

The Name Describes the Syndrome's Main Features

"Velo" - comes from the Latin word "velum" which means the palate and the back of the throat. In this syndrome it refers to either a cleft palate which may be complete, incomplete, or sub-mucous, or to the tone of the pharyngeal (throat) muscles which may be poor. Symptoms may include nasal regurgitation of feeds and speech which is delayed and/or hypernasal. The treatment involves speech therapy and surgery in most cases. VCFS is the most common syndrome associated with a cleft palate without a cleft lip, although a cleft lip may also occur.

"Cardio" refers to the heart. However, two thirds of children with VCFS have no heart problems. For those who do have a heart problem it can vary in severity from requiring surgery to minor abnormalities not requiring treatment. The main malformations are Tetralogy of Fallot, ventricular septal defect and right-sided aortic arch. Medical and technological advances have meant the survival rate of these children has improved dramatically in the last ten years. A heart ultrasound scan (echocardiogram) is often needed to determine if a congenital heart problem exists and to correctly assess the problem.

"Facial" - refers to similar facial features which may not be obvious in the early years of life. Children have the colouring and features of their

family but the face usually has low muscle tone and reduced movement. They may have a long nose with a prominent nasal bridge and small nostrils with deficient lateral prominences of the nose (aleque nasi). The jaw may be small, the cheek bones flat and the face may appear "long', The eyes may have almond-shaped eye margins and they may be long-sighted, needing spectacles.

The syndrome can be extraordinarily variable.

Other Features May Include:

Developmental delay when young and later learning difficulties varying from mild to severe.

Emotional and behavioural problems

Eye problems - small optic discs (eye nerves) tortuous retinal vessels, iris deficiencies and long sightedness. Eye examinations are recommended.

Gastro-intestinal problems such as feeding problems, gastroesophageal reflux, nasal regurgitation of feeds and constipation.

Hearing problems - eustachian tube dysfunction, middle ear infections and hearing loss. Hearing tests are recommended.

Hernias

Hypoparathyroidism - low levels of the parathyroid hormone characterised by low levels of calcium and high levels of phosphate in the blood, sometimes resulting in seizures.

Hypospadias - the opening of the urethra, for the passing of urine, is not correctly placed on the male genitals.

Immune system problems associated with T cells which help the body fight infection. (This has nothing to do with AIDS).

Laryngeal web - this is rare and usually diagnosed in the newborn as it can cause breathing problems.

Low muscle tone.

Microcephaly - small head size.

Microsomia - short stature and small build.

Scoliosis - lateral curvature of the spine.

Sprengel's shoulder - small shoulder blades positioned higher than normal and protruding, giving a web-shaped neck appearance.

Talipes - deformity of the foot.

Tapering fingers - get thinner towards the ends.

Undescended Testes - testes that have not descended into the scrotum.

Diagnosis

It is not uncommon for parents to have seen many doctors before the diagnosis is made. With further research and increasing awareness, this situation should improve over the next few years. Initial diagnosis is based on the recognition of the collection or pattern of features (as described above) in an individual that together constitute the syndrome.

Recognition is complicated by the many different features, their varying degrees of severity and the fact that some features appear to change according to the age. These changes are most commonly seen in the facial features.

Most cases have been identified through cleft palate and cranio-facial clinics and a few through cardiology, developmental, immunology and genetics services.

Children may be referred for genetics assessment and counselling. VCFS is non-progressive, meaning that the symptoms do not get progressively worse as the children get older. Research has revealed that, in the majority of children with VCFS, it is due to a submicroscopic absence of genetic material on the long arm of one chromosome 22. A special test to detect the absent genetic material on chromosome 22 is now available to confirm the diagnosis. In about 10-15% of cases it is inherited from a parent.

If parents have the special blood test for VCFS, and the change was detected in the number 22 chromosome, this test can be used in a future

pregnancy to check the fetus for VCFS. If the blood test was negative or has not been done, then an ultrasound scan can be performed at 18 weeks of pregnancy to check the baby's growth and development. However, the ultrasound is not very accurate for diagnosing VCFS.

Di George Syndrome

Dr Di George described features in a syndrome that are now known to be mostly due to the same deleted genes on chromosome 22 as VCFS. Di George syndrome features comprised a severe end of the spectrum of VCFS (or VCFS is the mild end of the spectrum of Di George). This variability can be intra familial - when inherited, a parent may have the "mild" VCFS and a child the "severe" Di George, or vice versa.

Is there a cure for VCFS?

No. Genetic conditions cannot be cured because faults in the genes cannot be fixed. Genetic research continues at a rapid rate but it is not appropriate to expect a cure for the condition as a whole. However, many of the features of VCFS can be improved or treated.

Treatment

This involves a thorough assessment of the child and treatment of the presenting problems, particularly speech problems. This may include surgery, speech therapy, physiotherapy, occupational therapy and remedial assistance at school. It is helpful for parents to have a general practitioner or paediatrician co-ordinate the various assessments and trips to health professionals involved with their child.

What is it like to have a child with VCFS?

When the diagnosis is made some parents are relieved to finally have answers, while others are saddened to know their child has a genetic condition. The grief which parents experience is natural and a part of coming to terms with their child having special needs. The parents may grieve when their child undergoes treatment such as surgery or are seen to be slightly different from their peers as they progress through the stages of life.

Parents often feel isolated because they do not know much about VCFS or know anyone else with an affected child. Professional agencies and the family doctor are often unaware of the associated problems because VCFS is a rare, only recently recognised condition.

Financial Support

Due to the extra parental time involved with caring for children with VCFS and helping them with their different types of therapy, many families are entitled to the Child Disability Allowance (CDA). The Department of Social Security then supplies a Health Care Card which enables the child to access health services and medications at minimal cost. This can be discussed with the General Practitioner.

The Velo-Cardio-Facial Syndrome Foundation of Australia.

This group consists of parents and professionals. The Foundation provides support for families through its meetings and social activities and information for families and professionals via its magazine.

The VCFS Foundation aims to:

* raise awareness of VCFS
* disseminate information and provide support to individuals with VCFS and their families.
* promote and support research in to VCFS and related disabilities
* promote and support education and development programs that will enhance the intellectual and social development of individuals with VCFS
* promote and support programs that will help individuals with VCFS achieve their maximum potential in the community.

For further information and references please contact:

Secretary VCFS FA

Mary Thorley

19 Eleanor Crescent

Rooty Hill, NSW 2766

Tel: (02) 625-3710

President,VCFSFA

Astrid King

38 Stanhope Rd

Killara NSW 2071

Tel: (02) 416-3759

FAMILY STORY

SIMON'S STORY

Simon was born in September 1989 after an uneventful pregnancy and normal delivery with a six hour labour. He is our second child. After two days it was noted he appeared dusky. On examination he had low blood gases and was transferred to Neonatal Intensive Care and incubated. He was treated for severe infection. After a cardiac consultation he was diagnosed with a congenital heart disorder that needed surgery within weeks. He had a right aortic arch and total anomalous pulmonary venous return. He came home on medication and weekly blood tests. I noticed him jittery and his GP tested for calcium levels. This was minimal and we were admitted to Kids Hospital Endocrine Ward for investigation. Simon needed calcium supplements. He had a cardiac catheterisation and was discharged. At six weeks he had open heart surgery for TAPVR repair and biopsy of thymus. He was diagnosed with Di George syndrome. At seven months he failed the hearing test at the Baby Clinic. On a subsequent visit we were referred to the National Acoustic Laboratories for hearing tests, these were inconclusive.

At eight months hehad his first bout of asthma.. Att eleven months he had a febrile convulsion with acute otitis media. We were referred to an ENT specialist who performed an echocohleagraph showing he need bilateral hearing aids. He was aided from sixteen months.

Simon crawled before he could sit and walked at fifteen months. He had no teeth till fifteen months. His height is under the third percentile and his paediatrician has always been concerned with this.

He was a very sickly child for the first two years, requiring weekly visits to the GP and associated specialists. He also had a few severe asthma attacks requiring hospitalization. He continues daily asthma treatments.

At two years he commenced early intervention for his deafness at St Gabriel's. This weekly program continued for three years. At three and half years Simon was diagnosed with Velo-Cardio-Facial syndrome (Shprintzen syndrome). This is a chromosomal abnormality with a deletion on chromosome 22. This came as quite a shock to us. Subsequent chromosomal studies in 1995 have shown he has the deletion. He also has the facial features, small aleque, long narrow nose, almond shaped palpebral fissures, prominent ears with infolded helix. He had an umbilical hernia and left inginal hernia (unrepaired). His teeth have required repair under anaesthetic. A history of two episodes of a collapsed lung and severe asthma delayed this till he was four and half years.

He has always been an overactive child and after some behaviour problems at a child care centre we were referred to a psychiatrist who diagnosed him with Attention Deficit Hyperactivity Disorder. After some drug trials he is now medicated every day. Simon is now six years old

and he attends kindergarten in the morning at St Gabriel's and the afternoon at his local school with his sister. He attends Occupational Therapy for fine motor exercises.

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BRIAN O'LEARY PLAYS CRICKET AND GOLF

The O'Leary's third child was diagnosed VCFS at age two-and-a-half during a first consultation with Dr Tony Lipson at the Children's Hospital, Camperdown. Brian O'Leary had a sub-mucous cleft palate which led parents Mick and Michelle to the cleft palate clinic at Camperdown, and the clinic referred them to Dr Lipson.

Soft palate repair was carried out a few weeks later, and a further operation at age seven has improved Brian's speech to a marked degree. The second operation proved a turning point, giving him a heightened self-confidence and a stronger will to achieve. Brian, now age nine, attends a composite class, years 4-5, at Primbee Primary School, just south of the NSW heavy industry town of Port Kembla where Brian's father is superintendent of transport for BHP, the predominant employer in this South Coast area.

Brian's diagnosis came as a relief to this mother Michelle, who has two older children, Matthew age thirteen, and Rebecca age eleven. After nearly three years of consulting doctors and paediatricians in an effort to determine the reason for their youngest son's slow development and speech problem, Mick and Michelle now had an identified syndrome for which the boundaries, if not the long term prognosis, are well charted. Dr Lipson suggested they get in touch with the parents of an older VCFS child to help them understand the implications and where the road might lead. That contact was made, and more contacts have been conducted over the years, all valuable courses of information. However, Mick and Michelle are aware of an occasional reluctance in other parents to get involved, a sensitivity which they understand, but feel is contrary to the best interests of the families with a VCFS child.

Mick is of the opinion that Brian is unaware he has a condition which makes it impossible to maintain learning parity with his peers. He is a strong verbal communication despite his twice-corrected palate giving a mild nasal intrusion. His good long sight allows him to wield a cricket bat with a fair degree of competence, and he swings cut-down ladies golf clubs to good effect. Significantly, Brian maintains easy friendships, and hasn't sustained any discriminatory treatment on the playground or the paddock.

While he is free of heart problems he does have other ongoing manifestations of the syndrome. Besides the nasal speech, his stature is small, remediation will be required to correct his sight for reading purposes, hearing is impaired and he has learning difficulties. Brian's physical appearance at age nine, is within the normal range. His parents took the decision to have his ears surgically corrected for comestic reasons. He continues to be a bed wetter, but the introduction of a waterbed has largely neutralised the inconvenience as well as reducing his allergic reaction to bed clothing. He does tend to be a follower, not a leader.

On the other side of the ledger, Brian has a strong will to succeed, and displays good physical co-ordination. He has a high tolerance to pain and never complains.

While he hasn't repeated any years at school, he does have reading difficulties, being about six months behind the norm, and mathematical concepts two years behind. His father calculates that the composite 4-5 class will provide an appropriate opportunity for his son to run through the fourth grade work again next year while retaining his mates in the same classroom as they move up to fifth grade work. Brian's desk is strategically located next to his teacher to whom he relates well, and this, allied to reading group participation and one hour a week private tutoring, has lifted his performance from a position of being unable to read two years ago to

the present day where he is behind by only six months. He continues with speech therapy, one attendance a fortnight.

What does the future hold for Brian? His parents are concerned that he may not be able to grasp mathematical concepts in high school, but are convinced he will achieve his full potential. The lack of scientific data on teenage VCFS children means they have to take one year at a time, but Mick and Michelle are encouraged by the accumulation of information coming from VCFS Foundation members and professionals operating in the field. Next year, Brian will take a place as a member of a school cricket team, a significant step in his development.

by

Len King

SARAH

OUR SPECIAL DAUGHTER

When Sarah was born we were told that she had a cleft palate which was repaired when she was 9 months old. Her first few years were probably the hardest.

She was a terrible feeder until after her cleft palate was repaired. We had to use a squeeze bottle as she was unable to suck properly. She was slow to put on weight and feeding times seemed to go on forever.

She has low muscle tone and did not walk until twenty one months. She seemed to suffer constant ear infections and had six grommets replacements in two half years. Her speech was slow to develop. I found it really strange as all her little friends seemed to babble constantly and our daughter was so quiet. We started speech therapy when Sarah was two and half and this has been on-going since then.

Our daughter was diagnosed with VCFS at the age of two half, although it was quite a shock it was almost a relief to know why our child did not seem to be developing at the same rate as all her friends and we were now able to address a specific problem.

Before she started pre-school at three and half we have several meetings with the school who had a special needs teacher who was able to work with our daughter during the morning activities. We were thrilled with the way she settled into her new environment; not once did we have a day when she did not want to go to school. The speech therapist, physio and special needs teacher all worked together to help improve her communication skills and mobility and we were able to prepare her successfully for school.

Our biggest concern now is how to address Sarah's learning difficulties and help her progress through the school system as easily as possible. Our school has been very supportive of her needs. It has taken persistence but she is now receiving extra support through the education system. At this stage she has been very keen to learn and we have wanted to capitalise on this as much as possible. Recently she has been diagnosed as long sighted and is now wearing glasses. As well, through Hans Peter Abel we are working on eye exercises and therapy to help improve her ability to focus and track to make reading easier.

Sarah is now seven and half and very special to us. You couldn't find a more caring and loving child. She has brought us so much joy and given us so much love that the battles we have had are all certainly worthwhile.

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