genetic alliance australia

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|| EDITORIAL

Welcome

The year 2020 has been challenging in new and different ways and I think we will all be glad to draw a line under 2020. I would like to thank everyone who wore masks, wash and sanitised their hands, went into lockdown and learnt how to use QR codes. We have learnt how to connect virtually, not only for work, but in our personal lives for family events, weddings and funerals. Geographical barriers have lessened as we have connected, but we all agree it is not the same as face to face.

Genetic Alliance held our first series of webinars for the metabolic condition, trimethylaminuria, TMAU. The viewings have exceeded our expectations and confirms this is as an excellent medium to share information with our community. We are planning more online information sessions for other conditions and will promote this through our social medial. Our team leader meetings have moved online with success and will continue in 2021.

Because of social distancing, we held our Genetic Disorders Awareness Week online with the theme of Genetics: Community, Connected and Digital. Thank you to all our presenters, MP Jenny Aitchison for sharing her personal story, Dr Alison McEwen on the UTS Master of Genetic Counselling course, and Dr Breda Cary for the Deaf and Hard of Hearing People. These presentations have subtitles and deaf signers for all the community to engage with this event.

As I write, it seems 2021 will bring similar challenges. I hope the new year break has refreshed you, our thoughts of community and was of being connected. May 2021 bring welcome challenges and much joy for you and your loved ones.

Best wishes and thank you for your ongoing support.
Jan Mumford
Executive Director

|| PROJECTS OFFICER

BY JENNY ROLLO OAM

How crazy was 2020? The whole year seemed to be challenging Murphy's Law: "Anything that can go wrong will go wrong".

I'm sure we could all add our own corollary or two. Notably: early in the year working from home, dealing with health issues when we were trying to avoid hospitals and spending the first ten minutes of every online meeting with "Can you hear me?", "Please mute yourself" and "Can everyone see the shared screen?".

At Genetic Alliance we have moved many of our meetings online. You can catch up with presentations through the News page of our web site.
Our program for leaders of condition-specific groups has actually been enhanced because of online meetings. Leaders can attend from anywhere in Australia, and presenters are able to share their wisdom from wherever they are based. We plan to continue this in future.
Where online meetings fail people is making those vital personal connections. We are hopeful that with the protection of a vaccine to enhance our public health initiatives we can soon start to ease restrictions. Will this happen in 2021? We'll have to wait and see. In the mean time we will have to be content with

webinars.
Wishing all our readers a very happy holiday season and good health in the coming year.

Be Kind to Yourself….
Jenny
Projects Manager

|| RARE DISEASE DAY 2020

Rare Disease Day 2020 is very rare indeed as this was a leap year, falling on Saturday 29th February. Genetic Alliance Australia partnered with SWAN and Sanofi. The evening was well attended with many families and portraits of their loved ones generously shared for all to view. Thank you to the speakers who generously spoke of their goals and developments and most significantly their experiences. These included Heather Renton, SWAN Australia; Nicole Millis, Rare Voices Australia; Lucinda Freeman, Genetic Counsellor; Agnes Snofwa, Australian Sickle Cell Advocacy and Jan Mumford, Genetic Alliance Australia. Shannae Ku spoke of her life with MPs. Guest Sam Humphrey, who performed in the Greatest Showman spoke of his life with skeletal dysplasia.

We were fortunate to be able to have this event just before the national lockdown.

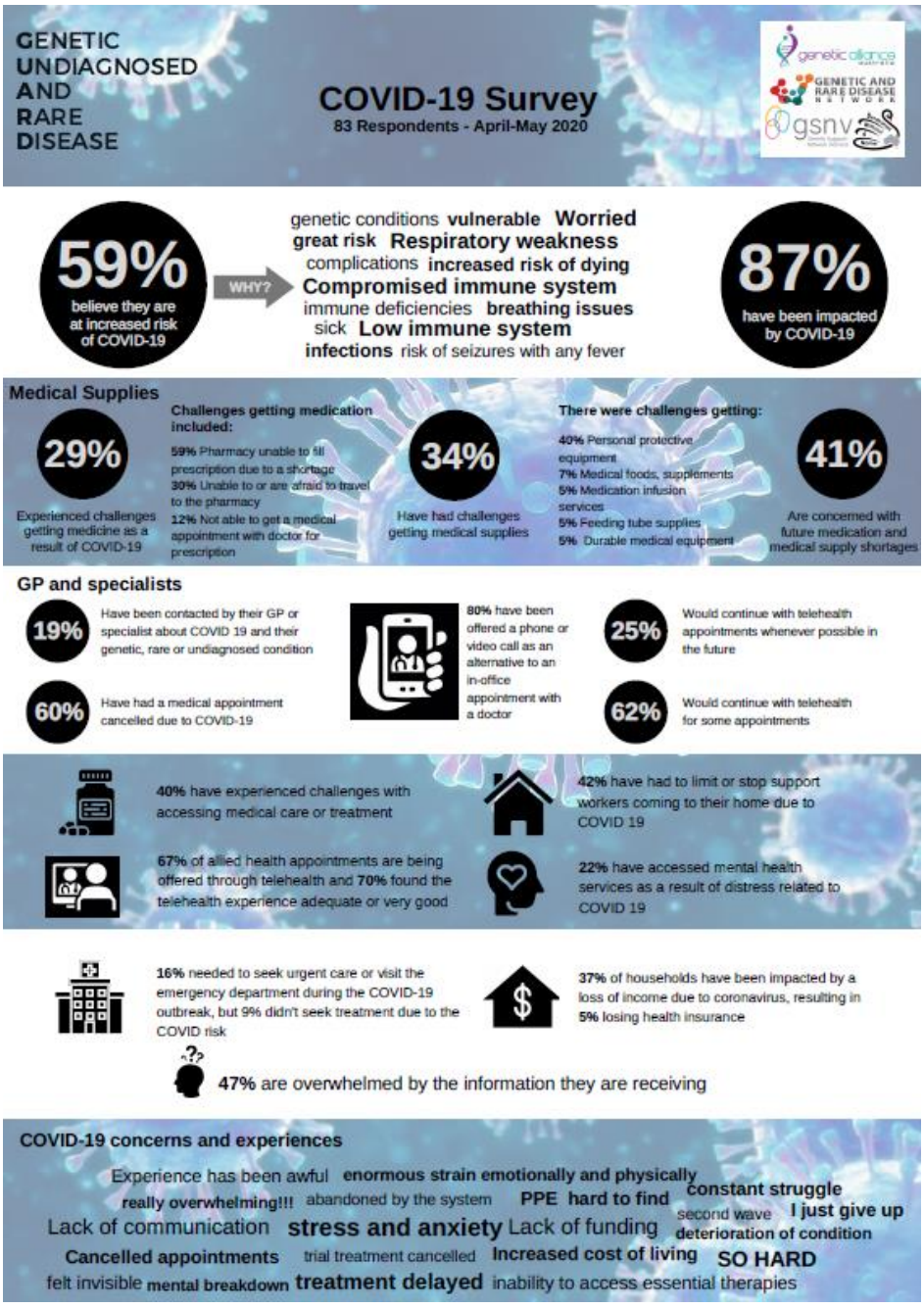
|| RARE DISEASE DAY 2021

Rare Disease Day 2021 will be very different with social distancing. Our event is in the planning stages. Pease watch social media for updates.

If you or your organisation are holding a function to mark this day please share your stories and photos with us! You can post your event on the Rare Disease Day web site here. Please send up your stories to ttps://www.rarediseaseday.org/send-us-your-event. To feature in our newsletter please email to director@geneticalliance.org.au or mail to Level 6 384 Victoria St Darlinghurst NSW 2010.

|| GENETIC UNDIAGNOSED AND RARE DISEASE COLLAORATIVE SURVEY

Following the initial lockdown to contain the pandemic, the GUARD Collaborative - Genetic Alliance, Genetic Service Network Victoria, Syndrome Without A Name and Genetic and Rare Disease Network conducted a survey on the impacts to our community. Thank you to everyone who completed this survey. This told us that a significant number 87% have been impacted by COVID-19. Many challenges were experienced with over one third having difficulties accessing medication, two thirds had appointments cancelled and 16% needed urgent care. They way health services were delivered changed as 80% were offered a pone or video consultation, with 62% saying they would continue with some appointments in this manner.



|| CHILDHOOD DEMENTIA INITIATIVE

> "Child dementia. The fact that these two words go together is appalling. We need to recognise this as a serious and urgent issue and fix it". Sean Murray Director Childhood Dementia Initiative.

Building on her experience with her children's diagnoses of San Filippo, and knowledge learnt founding the San Filippo Children's Foundation, Megan Donnell has founded the Childhood Dementia Initiative. Megan suspected childhood dementia as caused by a variety of genetic disorders, but could find no medical literature to confirm this. A white paper instigated found that childhood dementia is part of over 70 rare conditions, with more than 2 children diagnosed each week. There is economic costs and also the substantial impacts to family through grief, pain, anxiety, parents health and emotional toll.

For more information phone on 0414 482 541 or email hello@childhooddementia.org.

|| GENOMIC TESTING AVAILABLE ON MEDICARE

Long awaited, Medicare provision of genomic testing is now available. This overview describes some of the genetic or genomic testing available on Medicare from 1 May 2020.

This will be for some undiagnosed childhood syndromes or moderate to severe intellectual disability, in children 10 years old or younger. These genomic tests may be for the child or child and both biological parents depending on the Medicare item number. These tests need to be ordered by clinical geneticists or pediatricians, in consultation with clinical geneticists. For reference when discussing with your clinician the numbers are 73358, 73359, 73360, 73361, 73362, 73363.

The Centre for Genetics Education NSW Health has a series of 4 short educational videos to guide paediatricians on genomic testing. Viewed in sequence, the first video provides an introduction and overview of how to order genomic testing for the above Medicare items. The next 3 videos provide a step-by-step guide of what is involved including identifying suitable patients, choosing the appropriate genomic test, obtaining consent for the genomic test, explaining genomic test results and when to contact Genetic Services.

The Resource section, on the same webpage, contains fact sheets which explain genetic conditions or testing such as chromosomal microarray testing, Fragile X Syndrome, effect on life insurance, reproductive carrier screening and genetic/genomic testing.

Also released on 1 May 2020, are Medicare items (73354, 73355, 73356) for medical specialists to order testing for inherited gene variants with a higher chance of colorectal and endometrial cancer, such as those associated with causing Lynch Syndrome (LS), Familial Adenomatous Polyposis (FAP), MUTYH-Associated Polyposis (MAP), Juvenile Polyposis Syndrome (JPS), Peutz-Jeghers Syndrome (PJS) and Hereditary Mixed Polyposis

|| GENETIC AND RARE DISEASE TELEHEATLH NURSE CONSULTATION

Genetic Alliance has partnered with Centre for Community Driven Research to provide Telehealth Nurse consultations for those needing assistance in navigating our complex health system.

This is a free, confidential information and support service which provides information and links to practical support. You can book a time to talk with her by completing the online booking form. The Patient Pathways nurse can help you with any questions and/or concerns you may have including:

- emotional support if you or someone you care about has a genetic, undiagnosed or rare disease
- how to manage life with a genetic, undiagnosed or rare disease
- questions you may have about your, or the person you care for genetic, undiagnosed or rare disease
- how to find other organisations and resources that can help you or your family's life with a genetic or rare condition
- help on guiding you through the healthcare system to access all the relevant services.

We value your personal and health information and we will keep this confidential. For more details please see our privacy policy. Please note the Patient Pathways Nurse is only available to people living in Australia. Online bookings available on https://ccdr.knack.com/case-management#request-an-appointment, or call 1300 755 050

|| PERSONAL STORY

GATAXIA TELANGIECTASIA– HERE'S MY STORY

In 2019, Natalie Elkheir, Vice President of BrAsh A-T Ataxia Telangiectasia presented at GDAW event. For those who attended you may recall her presentation, printed below. Thank you Natalie for permission to tell your story.

It all started in 2007, when I made the most joyous discovery that I was pregnant. For my husband and I, this was extraordinary news as we had been trying for a baby for almost 10 years. I recall it was a time of excitement, anticipation and overwhelming jubilation! Finally this was happening, and we could not contain our delight.

Also in 2007, whilst we were experiencing the happiest time of our lives, another couple was experiencing the most devastating time of theirs. Sean and Krissy Roebig had received the news that their 2 youngest children, Brady and Ashleigh, had been diagnosed with the rare genetic children's condition called Ataxia Telangiectasia, or A-T for short. A-T is an autosomal recessive disorder primarily characterized by cerebellar degeneration, immunodeficiency, cancer susceptibility and radiation sensitivity. It is often referred to as a genome instability or DNA damage response syndrome. It is a progressive, degenerative, life-limiting and unrelenting condition affecting a variety of body systems with no known treatments or cure.

As they searched around desperately for support, for information, for treatments, they found nothing. And so they established a parent driven foundation committed to supporting families in Australia living with A-T, whilst funding vital research to find a cure. They named it BrAsh A-T in honour of their children, and their legacy continues today, 13 years later. Unbeknownst to me at the time, the Roebigs had laid the groundwork for what would become the greatest source of hope for me in years to come.

In April 2008, I welcomed into the world a beautiful little girl, Laila. And as she was placed in my arms for the first time, my hopes, my dreams and expectations for this newborn mini

me could only be described as infinite, and it was my job to ensure that she had the tools to achieve them all.

But as Laila grew, and failed to meet the expected milestones, it was becoming increasingly apparent that something was amiss. Sure, she couldn't hold her head up, but that will pass. OK, she looks like a drunken sailor when she walks, that will pass. Yes, yes, her speech is slurred, but that will pass. Only, it didn't pass. And so started the frustratingly long journey to discover why. We explored many genetic conditions, all of them rare, and each one seemingly worse than the last. But I was never convinced that any of them quite fit Laila's symptoms. Until one day, when Laila was 3.5 years old, I walked into an ophthalmologists office to assess whether her unsteadiness was due to issues with her vision. The doctor reported that actually, Laila's vision was fine...

HOWEVER, he said, ... she does have some red spider veins in her eyes called telangiectasia, which, coupled with her unsteady walking, may be an indication of something called Ataxia Telangiectasia. I thanked the good doctor for any direction he could give, and I bid him farewell.

That evening, I typed the words Ataxia telangiectasia into my google search bar. And as I scrolled through the information, immediately, I knew without any shadow of a doubt, that this was it. Laila had A-T.

The initial days and weeks following diagnosis were filled with profound despair and unbearable pain for my husband and I as we struggled to come to terms with the fate that awaited our little girl. No immunity .. lungs that may fail her ... loss of brain cells confining her to a wheelchair ... cancer ... may not survive beyond her teens. The fact that we were the source of her faulty genes, that we would have to watch our little girl, so vibrant and full of life, deteriorate before our eyes, and we could do nothing to prevent it was agonizing. We were told to take our little girl home, create memories, make every day count, for there was nothing that could be done to spare her from the ravages to come. The feelings of helplessness and hopelessness were crippling. A-T overshadowed everything, and every day was bittersweet.

But every day, oblivious to the fate that awaited her, Laila would wake, armed with an innocent smile and a sense of excitement for the day ahead. I soon realised that, just as the Roebigs had done before me, I had to make a choice … I had to choose whether to be defeated, or defiant. In making my choice, I felt empowered. I wanted to fight, I wanted to outrun A-T, and if A-T won the race, it wasn't because I hadn't tried everything in my power to beat it. A-T may take tomorrow, but I could not, and would not, allow it to deny us today.

And so was born my determination to making a difference to the outcomes for Laila and the 40 odd other known cases of A-T in Australia. However, I had a very limited understanding of the fundamentals of A-T, or genetics in general. I found myself wishing I had paid closer attention in biology at school. I had to become an expert on A-T, I had to push aside my insecurities, to engage people smarter than I and ask the dumb questions in order to gain an understanding of just what was going on inside every cell of Laila's body. And before long, terms like chromosomes and exons, DNA, RNA and amino acids, become part of my vocabulary.

Armed with some knowledge of what A-T was, my husband and I engaged our family, friends and community with several fundraising campaigns to support BrAshA-T fund vital research in the hope of furthering our understanding of this genetic minefield. Despite early success with this approach, we soon realised that we could not continue to tap into the pockets of our limited network, that the funding needed to make significant progress was far more than altruism alone could achieve.

With the added challenge of A-T being a rare genetic disorder, engaging other affected families meant that we only had a group of about 40 others to call upon. These families were at differing stages of the condition, many had resigned themselves to the fact that their child had already deteriorated beyond the scope of any potential treatment, and their focus was centred on the care and happiness of their family. Others struggled with the load of the increasing needs of their child and had little time to give to the cause. But all were doing the best they could, and it was cruel and unfair to expect them to contribute whilst they struggled to simply survive.

BrAshA-T's founding family, the Roebigs, were also not immune from this crippling burnout. In a cruel twist of fate, Sean Roebig, as a carrier of

an A-T mutation, was also susceptible to cancer, as all carriers are. He was diagnosed with melanoma and sadly succumbed to its effects after it had metastasised. Widowed, and with her children's needs requiring greater attention, Krissy was under extreme strain, detracting from her ability to spearhead the campaign for a cure she had started years before.

And so this organisation that was driving change and progress from the ground up, faltered due to the harsh reality of A-T which appeared to be leaving a trail of devastation wherever I looked. It was at this point that I decided to extend my support for BrAshA-T beyond just fundraising, and join the board of directors.

Today, as I serve as Vice President of BrAshA-T, I am so thankful for the legacy left by the Roebig's and I am proud of the advances we have since made in our fight for a cure. Initiatives such as funding a National A-T Clinic, accessible to all A-T patients in Australia, this alone has seen a significant improvement in the health outcomes of our patient base.

However, with such a small A-T community in Australia, it was apparent that collaboration with other A-T organisations was imperative to accelerating research. I therefore travelled to international A-T clinical conferences to make these connections and had great success. By joining forces with parent driven organisations in the US, UK and other European countries, we have ensured that we are not doubling up on research, and that we collectively identify, with the support of our respective scientific advisory committees, the most promising research opportunities, stretching our limited funding even further. The advancement in our understanding of A-T since Laila was diagnosed 8 years ago has been incredible. We are on the verge of a cure for A-T, it is no longer a matter of if, but when. This progress was only made possible by the sheer determination of parents from around the world, unwilling to be at the mercy of for-profit research, sharing 1 common goal - to save their children. This is the changing dynamic driving genetic research.

Today, as I struggle to watch my daughter deteriorate, having once marvelled at her determination to be a junior cheerleader for her favourite footy team, while now watching her living her life on the periphery, going largely unseen and unheard as her world gets increasingly smaller, I fear the time will come when I, too, must pass the baton on to another mother, or father, motivated by despair to save their child from A-T, confident in the knowledge that there is no greater force to be reckoned with.

|| PROFILE

A-Z OF GENETIC CONDITIONS

ATAXIA TELANGIECTASIA

Synonyms of Ataxia Telangiectasia

- AT
- Cerebello-Oculocutaneous Telangiectasia
- Immunodeficiency with Ataxia Telangiectasia
- Louis-Bar Syndrome

General Discussion

Ataxia telangiectasia (AT) is a complex genetic neurodegenerative disorder that may become apparent during infancy or early childhood. The disorder is characterized by progressively impaired coordination of voluntary movements (ataxia), the development of reddish lesions of the skin and mucous membranes due to permanent widening of groups of blood vessels (telangiectasia), and impaired functioning of the immune system (i.e., cellular and humoral immunodeficiency), resulting in increased susceptibility to upper and lower respiratory infections (sinopulmonary infections). Individuals with AT also have an increased risk of developing certain malignancies, particularly of the lymphatic system (lymphomas), the blood-forming organs (e.g., leukemia), and the brain.

In those with AT, progressive ataxia typically develops during infancy and may initially be characterized by abnormal swaying of the head and trunk. As the disease progresses, the condition leads to an inability to walk (ambulation) by late childhood or adolescence. Ataxia is often accompanied by difficulty speaking (dysarthria), drooling; and an impaired ability to coordinate certain eye movements (oculomotor apraxia), including the occurrence of involuntary, rapid, rhythmic motions (oscillations) of the eyes while attempting to focus upon certain objects (fixation nystagmus). Affected children may also develop an unusually stooped posture and irregular, rapid, jerky movements that may occur in association with relatively slow, writhing motions (choreoathetosis). In addition, telangiectasias may develop by mid-childhood, often appearing on sun-exposed areas of the skin, such as the bridge of the nose, the ears, and certain regions of the extremities, as well as the mucous membranes of the eyes (conjunctiva).

AT is inherited as an autosomal recessive trait. The disorder is caused by changes (mutations) of a gene known as ATM (for "AT mutated") that has been mapped to the long arm (q) of chromosome 11 (11q22.3). The ATM gene controls (encodes for) the production of an enzyme that plays a role in regulating cell division following DNA damage.

Signs & Symptoms

An early symptom of ataxia telangiectasia is diminished muscle coordination usually noticed when a child begins to walk. Coordination (especially in the head and neck area) becomes impaired, and tremors (involuntary muscle contractions) can occur. In most cases, mental functioning is not affected and most children exhibit normal or above average intelligence.

The telangiectasias (visible dilated blood vessels) usually begin in the eyes (the eyes look “bloodshot”) between three and six years of age, although they can occur earlier. These discolorations may spread to the eyelids, face, ears, roof of the mouth and possibly other areas of the body. Rapid eye blinking and movements, and turning of the head may develop gradually. Occasional nosebleeds may also occur. The adenoids, tonsils and peripheral lymph nodes may develop abnormally or fail to develop. Muscle coordination in the head and neck area may be gradually impaired causing poor cough reflexes and problems with swallowing, breathing, drooling, and choking. Slurred speech and variable jerking, writhing and tic-like movements also be noticed.

Growth retardation can be linked to a growth hormone deficiency. Premature aging occurs in approximately ninety percent of affected individuals and is characterized by gray hair with dry, thin, wrinkled or discolored skin during adolescence. A variety of other skin or hair problems may develop in some cases. Abnormalities of hormone producing (endocrine) glands may be accompanied by incomplete sexual development in both males and females.

Because of an impaired immune response, affected individuals may be more susceptible to chronic sinus and/or lung infections, recurring cases of pneumonia and chronic bronchitis.

Persons with this disorder may be affected by a high incidence of carcinoma and lymphoma usually beginning during early adulthood. Approximately one in three affected individuals develop cancer, usually cancer of certain malignancies, particularly of the lymphatic system (lymphomas) or of the blood (leukemia). Exposure to x-rays seems to increase the incidence of possible tumors. In addition, individuals with one ataxia telangiectasia gene (carriers) also appear to have an elevated risk for cancer. Close relatives of persons with ataxia telangiectasia may be at a higher risk of developing certain types of cancer than the general population.

In some cases, a mild form of diabetes mellitus may occur. Diabetes mellitus is a condition in which there is insufficient secretion of the hormone insulin. Primary symptoms may include abnormally increased thirst and urination (polydipsia and polyuria), weight loss, lack of appetite, and fatigue.

Causes

Ataxia telangiectasia is inherited as an autosomal recessive trait. Genetic diseases are determined by two genes, one received from the father and one from the mother.

Recessive genetic disorders occur when an individual inherits the same abnormal gene for the same trait from each parent. If an individual receives one normal gene and one gene for the disease, the person will be a carrier for the disease, but usually will not show symptoms. The risk for two carrier parents to both pass the defective gene and, therefore, have an affected child is 25% with each pregnancy. The risk to have a child who is a carrier like the parents is 50% with each pregnancy. The chance for a child to receive normal genes from both parents and be genetically normal for that particular trait is 25%.

The disease gene that causes ataxia telangiectasia, known as the ATM gene, is located on the long arm (q) of chromosome 11 (11q22.3). Chromosomes are found in the nucleus of all body cells. They carry the genetic characteristics of each individual. Pairs of human chromosomes are numbered from 1 through 22, with an unequal 23rd pair of X and Y chromosomes for males and two X chromosomes for females. Each chromosome has a short arm designated as "p" and a long arm identified by the letter "q". Chromosomes are further subdivided into bands that are numbered.

Researchers have determined that the ATM gene encodes a protein that plays a role in regulating cell division after DNA damage. (DNA or deoxyribonucleic acid is the carrier of the genetic code.) The protein, which is known as ATM for "A-T mutated", is an enzyme (protein kinase) that normally responds to DNA damage by triggering the accumulation of a protein (p53) that prevents cells from dividing (tumor suppressor protein). However, in individuals with ataxia telangiectasia, abnormal changes (mutations) of the ATM gene cause an absence or defect of the ATM protein and delayed accumulation of the p53 protein. As a result, cells with DNA damage continue dividing (replicating) without appropriate repair of their DNA, causing an increased risk of cancer development. Approximately half of human cancers are thought to be characterized by abnormalities affecting the activity of the p53 tumor suppressor protein. Exposure to ionizing radiation (such as x-rays) normally enhances the p53-directed activity of the ATM protein; however, in individuals with ataxia telangectasia, deficient activity of the ATM protein results in extreme sensitivity to such radiation.

Affected Populations

Ataxia telangiectasia usually begins during infancy (between one and three years of age) and often affects more than one child in a family. Males and females may be affected in equal numbers. In the United States, the prevalence is approximately one in 40,000-100,000 live births.

Related Disorders

Ataxia means walking with an unsteady gait caused by the failure of muscular coordination or irregularity of muscular action. There are many forms of ataxia. Some ataxias are hereditary, some have other causes and sometimes ataxia can be a symptom of other disorders. To locate information about other types of ataxia choose "ataxia" as your search term on the Rare Disease Database.

Symptoms of the following disorders can be similar to ataxia telangiectasia. Comparisons may be useful for a differential diagnosis:

Friedreich's ataxia is a genetic, progressive, neurologic movement disorder that typically becomes apparent before adolescence. Initial symptoms may include unsteady posture, frequent falling, and progressive difficulties walking due to an impaired ability to coordinate voluntary movements (ataxia). Affected individuals may also develop abnormalities of certain reflexes; characteristic foot deformities; increasing incoordination of the arms and hands; slurred speech (dysarthria); and rapid, involuntary eye movements (nystagmus). Friedreich's ataxia may also be associated with cardiomyopathy, a disease of cardiac muscle that may be characterized by shortness of breath upon exertion (dyspnea), chest pain, and irregularities in heart rhythm (cardiac arrythmias). Some affected individuals may also develop diabetes mellitus, a condition in which there is insufficient secretion of the hormone insulin. Primary symptoms may include abnormally increased thirst and urination (polydipsia and polyuria), weight loss, lack of appetite, fatigue, and blurred vision. Friedreich's ataxia may be inherited as an autosomal recessive trait. (For more information on this disorder, choose "Friedreich's ataxia" as your search term in the Rare Disease Database.)

Marie's ataxia is a neuromuscular syndrome inherited as a dominant trait. Also known as Pierre Marie's disease or hereditary cerebellar ataxia, it often begins during the third or fourth decade. An early symptom is unsteadiness walking down stairs or on uneven ground. Frequent falls may occur as the disorder progresses as well as tremors, loss of coordination in the arms and speech disturbances. In later stages slight loss of vision, and loss of pain or touch sensations, may also occur. Swallowing and clearing of secretions may eventually become difficult if the throat muscles are affected. (For more information on this disorder, choose "Marie" as your search term in the Rare Disease Database.)

Charcot Marie Tooth hereditary neuropathies are a group of disorders in which the motor and sensory peripheral nerves are affected, resulting in muscle weakness and atrophy, primarily in the legs and sometimes in the hands. CMT hereditary neuropathy affects the nerves that control many muscles in the body. The nerve cells in individuals with this disorder are not able to send electrical signals properly because of abnormalities in the nerve axon or abnormalities in the insulation (myelin) around the axon. Specific gene mutations are responsible for the abnormal function of the peripheral nerves. Charcot Marie Tooth hereditary neuropathy can be inherited in an autosomal dominant, autosomal recessive or X-linked mode of inheritance. (For more information on this disorder, choose "CMT" as your search term in the Rare Disease Database.)

Hereditary olivopontocerebellar atrophy (OPCA) is a rare group of disorders characterized by progressive balance problems (disequilibrium), progressive impairment of the ability to coordinate voluntary movements (cerebellar ataxia), and difficulty speaking or slurred speech (dysarthria). There are at least five distinct forms of hereditary OPCA. All forms of hereditary OPCA, except one, are inherited as autosomal dominant traits. The term olivopontocerebellar atrophy has generated significant controversy and confusion in the medical literature because of its association with two distinct groups of disorders, specifically multiple system atrophy (MSA) and spinocerebellar ataxia (SCA). OPCA may refer to a specific

form of MSA or one of several types of SCA. Hereditary OPCA refers to the group of disorders that overlaps with SCA. Both forms of OPCA are characterized by progressive degeneration of certain structures of the brain, especially the cerebellum, pons, and inferior olives. The cerebellum is the part of the brain that plays a role in maintaining balance and posture as well as coordinating voluntary movement. The pons is part of the brainstem and contains important neuronal pathways between the cerebrum, spinal cord, and cerebellum. The pons serves as a relay point for messages between these structures. The inferior olives are two round structures that contain nuclei that are involved with balance, coordination and motor activity. (For more information on this disorder, choose "hereditary olivopontocerebellar atrophy" as your search term in the Rare Disease Database.)

Diagnosis

A diagnosis of ataxia telangiectasia is made based upon a detailed patient history, a thorough clinical evaluation, identification of characteristic symptoms, and a variety of specialized tests including blood tests, magnetic resonance imaging (MRI), and karyotyping.

Blood tests may detect elevated levels of serum alpha-fetoprotein, which occurs in approximately 85 percent of cases. However, in unaffected children this protein may remain elevated until 2 years of age. Blood tests may also reveal elevated liver enzymes. During an MRI, magnetic field and radio waves are used to create cross-sectional images of the brain, which can show progressive cerebellar atrophy. Karyotyping is a specialized test that detects chromosomal abnormalities. Affected individuals have an increased frequency of such chromosomal abnormalities.

Standard Therapies

Treatment

Treatment for AT is directed toward control of symptoms. For respiratory infections, therapy with an antibiotic drug, postural drainage (with the head lower than the rest of the body) of the bronchial tubes and lungs, and gammaglobulin injections in some cases may be effective.

Avoidance of undue exposure to sunlight may help control spread and severity of dilated blood vessels (telangiectasias). Vitamin E therapy has in some cases been reported to provide temporary relief of some symptoms, but should only be tried under advice and supervision of a physician to avoid toxicity. The drug Diazepam (Valium) may be useful in some cases to help slurred speech and involuntary muscle contractions. Physical therapy may help maintain muscle strength and prevent limb contractures. Care should be taken to ward off infections.

Other treatment is symptomatic and supportive. Genetic counseling may be of benefit to persons with AT and their families.

Investigational Therapies

Information on current clinical trials in Australia is posted on the Internet at www.clinicaltrials.gov.au
Information kindly provided by NORD on https://raredisease org/rare-diseases/ataxia-telangiectasia/

|| CONTACT CORNER

Genetic Alliance has received request for contact from people searching for families or individuals with similar conditions to share information and experiences.

Acrodermatitis enteropathica
Potocki Shaffer syndrome
McCune-Albright syndrome
Velo cardio facial syndrome
Wegener's granulomatosis

Orbital myositis (also known as Ocular myositis

Please email info@geneticalliance.org.au or phone on 02 9295 8359 if you would like to make contact with these enquiries or for other conditions.

|| SUPPORT GROUP MEETINGS CONFERENCES & RESEARCH

Due to social distancing support group meetings have been postponed. Some organisations have moved to virtual meetings. Please contact your support group leaders for more details.

|| MEMBERSHIP AND DONATIONS

Due to the pandemic and the impacts to families Genetic Alliance Australia Board decided to suspend membership fees for 2020-2021. This will be reassessed in mid 2021.

Genetic Alliance Australia is a small and vital not-for profit assisting those affected by rare genetic conditions, family, loved ones and community. To keep up to date with happenings, please join and becoming a member and receive free electronic updates. To be transferred to the electronic mail system, please email us on admin@genticalliance.org.au or phone on 92958365.

If you prefer a printed copy of this newsletter in the mail, we will continue this service. If you are new to this newsletter and would like to receive a printed copy, please email us on admin@genticalliance.org.au or phone on 92958365. Alternatively subscribe through our webpage. http://www.geneticalliance.org.au/membership.php

You may wish to give a donation – thank you! This will help in our independence and providing services in the future. All donations over $2 are tax deductible. Please donate here

https://www.paypal.com/donate/token=FiAjMFvTrGWzGPllCOtSO4bUVC_REdPoDHquIAAuNuWjROFFnQCq0E7dPvjg0vpSWI0OOG&country.x=AU&locale.x=AU

|| HEALTHCARE VISTING GUIDE: ENABLING FAMILY PRESENCE IN A COVID NORMAL WORLD

Genetic Alliance Australia worked with Health Consumers NSW to prepare the Healthcare Visiting Guide: enabling family presence in a COVID normal world.

This publication was developed with the *COVID-19 Consumer Leaders Taskforce* and the *COVID-19: Consumer Representative Hub* on Amplify. Laila Hallam, Chair of the Consumers Leaders Taskforce, and Serena Joyner, HCNSW Consumer Engagement Manager, were instrumental in bringing this together.

Since COVID-19 began there has been a tension between maintaining infection control in hospitals and allowing families to visit. Many patients and family feel that services have not always got the balance right.

This guide seeks to support effective family presence policies and practices through:

1. Recognising and accepting family presence as the accepted norm, while minimising the risk of infection introduction and spread
2. Explore new and innovative ideas and solutions with patients and families, as partners
3. Plan as a system; apply locally; and review on facts as the situation changes
4. Restore and revitalise stakeholder confidence – 'Together, we've got this!'

A copy of this guide is available at hhttps://amplify.hcnsw.org.au/visiting-guide .Or contact the office on 02 92958356 for a copy.

|| GOVERNT FEEDBACK AND SUBMISSIONS

Genetic Alliance Australia with the GUARD partnership, Syndrome Without A Name, SWAN Australia, Genetic Service Network Victoria and Genetic and Rare Disease Network WA, GaRDN provided feedback on two areas of interest. One was for the consultation on the Medical Services Advisory Committee Revised Draft Guidelines. The other was the Standing Committee on Health, Aged Care and Sport: Inquiry into approval processes for new drugs and novel medical technologies in Australia.

The first submission focused on the use of 'personal utility', or the value of knowing. In health and genetics 'knowing' your condition provides us with a sense self awareness, direction in our lives and helps communication with clinicians in decisions regarding treatments and outcomes. GUARD supported the use of personal utility and the power of knowing. This was not previously included in the guidelines.

The second submission called on the need for a more rapid approval process for new drugs and technologies. For those with rare disease, Australia is lagging behind in legislation, a difficult and burdensome approval process and there is no unified approach to achieving equity across the health needs of all of our people, including those with rare conditions.

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Monday to Friday: 9am – 5pm

Genetic Alliance Australia aims to:-

Provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group.

Facilitate access 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.

Educate the medical and allied health professionals and the community about genetic disorders.

Consult with government bodies, both Federal and
State, for appropriate funding for genetic services.

Prof. R.Trent PhD; BSC (Med); MB BS (Syd; BPhil (Oxon), FRACP; FRCPA.

Prof B Wilcken MB;ChB;FRACP

Subscription Year - 1 July to 30 June

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