

“JABE BABE – A HEIGHTENED LIFE”

52 minute documentary by Janet Merewether

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Distribution and Sales

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EDUCATION RESOURCES

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BACKGROUND INFORMATION - MARFAN SYNDROME

- Marfan Syndrome, a connective tissue disorder, is one of the most common but least publicly known genetic illnesses, affecting around 1 in 3000 people.

Common genetic disorders and their (approximate) incidence:

Down Syndrome	1 in 1100
Cystic Fibrosis	1 in 2,500
Marfan Syndrome	1 in 5,000

- Marfan Syndrome is caused by a mutation of the Fibrillin gene FBN1 on chromosome 15
- Marfan Syndrome can be spontaneous or inherited - 20% are mutations, but it is more common to inherit it.
- Prior to cardio-vascular surgery, the average age of death was 32 years, often the result of aortic rupture.
- Skeletal abnormalities include a long and slender stature, chest deformities (pectus excavatum), arachnodactyly or 'spider fingers', excessive length of arm, hyperextensible joints, and a high arched palate. Other features of the disorder may include lens and retinal detachment, foot deformities and heart problems, especially the dilatation of the ascending aorta.
- Though mostly hereditary, many people suffering from Marfan Syndrome are unaware of their condition, and are at risk of early death if left undiagnosed.
- Geneticist Dr Lesley Adès at The Children's Hospital at Westmead in Sydney has just developed the world's first effective blood test to identify the mutation of the Fibrillin gene on chromosome 15 in individuals and families. Today, with increased expertise, the condition can be managed, but Marfan patients live with the knowledge that their hearts & bodies are particularly vulnerable to problems such as aortic aneurism, blood clots and haemorrhage, even if the condition is already diagnosed, and the patient is taking blood thinning drugs such as Warfarin.
- If women with the Marfan Syndrome choose to have children, they both risk their own health, as well as their offspring, who have a 50% chance of inheriting the disease. Like many other genetic illnesses, the ethical debate about genetic

testing for 'imperfections' is contentious. Genetic testing for Marfan Syndrome is available and can be offered early in pregnancy or prior to conception by pre-implantation genetic diagnosis.

The Children's Hospital at Westmead Sydney offers blood testing to determine whether a person, or potential mother, has Marfan Syndrome. The hospital also has counselling services for families of Marfan sufferers, with doctors such as Dr Lesley Adès in Clinical Genetics taking a special interest in the condition.

- Marfan Syndrome has been publicized in sport magazines because of the sudden, untimely deaths of Olympic volleyball player Flo Hyman and collegiate basketball player Chris Patton who had the condition.
- In the USA, the NBA is considering testing for the syndrome as basketballers with undiagnosed Marfan Syndrome have dropped dead on the court.
- Abe Lincoln has been the subject of a successful court case to test his DNA for Marfan Syndrome. Other well known people with the condition include the violinist Paganini and the Egyptian pharaoh Akhenaten. There has also been speculation as to whether Osama Bin Laden suffered from the condition.

PHYSICAL MANIFESTATION OF MARFAN SYNDROME

BODY

- Longitudinal growth is everywhere, resulting in a high arched palate, long narrow face, eye problems and long limbs. Marfan sufferers are said to resemble portraits by El Greco.
- Long and slender stature
- Pronounced stretch marks on the shoulders, hips and lower back
- Chest deformities Pectus Carinatum or 'pigeon chest' and Pectus Excavatum (the opposite of pigeon chest)
- Excessive length of limbs
- Arachnodactyly – 'spider fingers'
- Long, extremely narrow feet and toes
- Scoliosis
- Hyper-mobile joints (can pull thumb back etc, double jointed)

EYES

- Eye problems in 50 – 70% of people
- Ectopia Lentis – lens detachment
- Tremors of the iris, cataracts and severe myopia and possible retinal detachment

HEART

- Mitral valve prolapse occurs in 75 – 85% of people and only occurs in 5% of the general population
- Prolapse can only be detected by an ECG
- Mitral valve regurgitation, i.e. a leaking valve (33%)
- Aortic valve is affected – stress from the beating heart and the lack of elasticity dilates the valve and often results in an aortic aneurism

- Basically an overworked heart
- Aortic dissection, when the aorta tears
- Repair to the heart is a composite dacron graft requiring open heart surgery procedure.
- Requires blood thinning medication to prevent blood clots and risk of haemorrhage
- Prone to infection of the valve (endocarditis), which often requires Marfan patients to be hospitalised for minor procedures.

LIST OF SELECTED AUSTRALIAN SUPPORT SERVICES AVAILABLE IN THE AREAS OF GENETIC RESEARCH/CONSELLING, MARFAN SYNDROME, SCHIZOPHRENIA, FOSTER CARE, CHILD ABUSE/NEGLECT.

MARFAN SUPPORT ORGANISATIONS

Marfan Foundation - NATIONAL

<https://marfan.org.au>

Genetic Alliance Australia

https://www.geneticalliance.org.au/conditions_detail.php?Marfan-syndrome-341

Health Direct - NATIONAL

<https://www.healthdirect.gov.au/marfan-syndrome>

CLINICAL GENETICS + MEDICAL SUPPORT SERVICES

List of National Genetic Clinics

<https://www.genetics.edu.au/genetic-services/general-genetics-clinics#ACT%20GC>

The Centre for Genetics Education - NSW

<https://www.genetics.edu.au/individuals-and-families/genetic-conditions>

(counselling and information)

Children's Hospital at Westmead - NSW

Department of Clinical Genetics

<https://www.schn.health.nsw.gov.au/find-a-service/health-medical-services/clinical-genetics/chw>

Victorian Clinical Genetics Services - VIC

<https://www.vcgs.org.au>

South Australian Clinical Genetics Service - SA

Women's and Children's Hospital , North Adelaide

<https://www.wch.sa.gov.au>

Genetic Service of Western Australia - WA

https://ww2.health.wa.gov.au/Articles/F_I/Genetic-Services-of-WA

Genetic Health Queensland - QLD

<https://metronorth.health.qld.gov.au/rbwh/genetic-health-queensland>

Tasmanian Clinical Genetics Service - TAS
<https://www.health.tas.gov.au/health-topics/genetics/genetics-service>

INTERNATIONAL GENETIC / MARFAN SUPPORT ORGANIZATIONS

Genetic Aortic Disorders Association, Canada
<http://www.gadacanada.ca/marfan-syndrome> CANADA

The Marfan Foundation
<https://marfan.org> USA

Marfan Trust
<https://www.marfantrust.org> UK

Marfan World
Formerly International Federation of Marfan Syndrome
Organisations (IFMSO)
<http://www.marfanworld.org>

National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/marfan-syndrome/>

Mendelian Inheritance in Man On-line (OMIM)
<https://www.omim.org/about>

OUT-OF-HOME & FOSTER CARE INFORMATION / CRISIS SERVICES

AFCA Australian Foster Care Association
<https://www.fostercare.org.au> NATIONAL

ACWA – The Association of Children’s Welfare Agencies
<https://www.acwa.asn.au/about/> NATIONAL

White Ribbon Australia
<https://www.whiteribbon.org.au/About-Us/Our-Vision> NATIONAL

FACS (Department of Family and Community Services)
<https://www.facs.nsw.gov.au> NSW

ACYP NSW Advocate for Children and Young People
<https://www.acyp.nsw.gov.au> NSW

Ombudsman NSW
<https://www.ombo.nsw.gov.au/what-we-do/about-us/who-we-are> NSW

Children's guardians and other offices

Office of the Children's Guardian - New South Wales
<https://ocg.nsw.gov.au> NSW

Guardian for children and young people – South Australia https://gcyp.sa.gov.au/what-we-do/your-rights-in-care/	SA
Commission for Children and Young People https://ccyp.vic.gov.au/resources/useful-links/	VIC
OPG - Office of the Public Guardian, Queensland https://www.publicguardian.qld.gov.au/about-us/our-purpose/child-advocate	QLD
ACT Human Rights Commission – Public Advocate https://hrc.act.gov.au/public-advocate/	ACT
<u>HELPLINES – CHILDREN IN CRISIS</u>	
Kids Helpline https://kidshelpline.com.au Tel: 1800 55 1800	NATIONAL
Lifeline Tel: 13 11 14fa https://www.lifeline.org.au	NATIONAL
Community Services Child Protection Helpline 132 111 https://www.health.nsw.gov.au/parvan/childprotect/Pages/default.aspx	NSW
Department for Communities – Child Protection tel: 1800 273 889 https://www.wa.gov.au/organisation/department-of-communities/child-protection	WA
Community services – Children and Families Child Abuse or Neglect helpline Tel: 1300 556 729 https://www.communityservices.act.gov.au/ocyfs	ACT
NT Families Housing and Communities Tel: 1800 700 250 https://tfhc.nt.gov.au/contacts	NT
Dept Children, Youth Justice, Multicultural Affairs Reporting Child Abuse Tel: 1800 177 135 https://www.cyjma.qld.gov.au/protecting-children/reporting-child-abuse	QLD
Dept Child Protection Child Abuse or Neglect Report Line Tel: 13 14 78 http://childprotection.sa.gov.au	SA
Department of Health	TAS

Tel: 1800 000 123
<https://www.health.tas.gov.au/contact-us/emergency-contacts>
<https://strongfamiliesafekids.tas.gov.au>

Dept. of Families, Fairness and Housing
After hours Child Protection Services
Tel: 13 12 78
<https://services.dffh.vic.gov.au/child-protection-contacts> VIC

ADDITIONAL INFORMATION – CHILD ABUSE/NEGLECT

Australian Government
Australian Institute of Family Studies
<https://aifs.gov.au> NATIONAL

Create Foundation
Consultants, educators and advocacy group for children in
out-of-home care
Helpline: 1800 655 105
create@create.org.au
<https://create.org.au/contact/>

Pathways of Care Longitudinal Study: outcomes of children
and young people in out-of-home care
<https://apo.org.au/node/314073>

See also
National Child Protection Clearing House
Senate inquiry into Children in Institutional Care

NSW Dept of Communities and Justice – Keep them Safe
Wood Special Commission of Inquiry
<https://theirfuturesmatter.nsw.gov.au/about-us/other-reforms/keep-them-safe> NSW

SCHIZOPHRENIA and MENTAL HEALTH SERVICES

SANE Australia
Helpline: 1800 18 7263
<https://www.sane.org/information-stories/facts-and-guides/schizophrenia> NATIONAL

Mental Health Foundation Australia
Tel: 1300 643 287
<https://www.mhfa.org.au/CMS/about-mhfa> NATIONAL

Mental Illness Fellowship of Australia (MIFA)
Tel: 1800 985 944
<https://www.mifa.org.au/en/> NATIONAL

One Door Mental Health
Formerly Schizophrenia Fellowship of NSW
Helpline: 1800 843 539 NSW

<https://www.onedoor.org.au>

see also fellowships in different states, such as
Mental Illness Fellowship of Victoria

SEXUAL ASSAULT SERVICES

NSW Health Sexual Assault Services

NSW

<https://www.health.nsw.gov.au/parvan/sexualassault/Pages/health-sas-services.aspx>

Child Protection Unit – Children’s Hospital at Westmead

Daytime: (02) 9845 2434

After Hours: (02) 9845 0000

see also services in each state