



**Muscular
Dystrophy WA**

Meet a Member... Jakob R

My name is Jakob I am 23 and have Duchenne muscular dystrophy (DMD). I was diagnosed at the age of 8; which is considered relatively late for persons with my condition. I require an electric wheelchair for daily mobility, as DMD greatly effects my leg and arm movement.

Recently I graduated from UWA with a Bachelor of Arts in Communication and Media Studies, and Anthropology and Sociology. The whole university experience was very enlightening, one which taught me fundamental writing and research skills. As a person with DMD, completing a degree is a significant achievement when you consider the additional challenges you are confronted with, and the adaptations which are required to successfully finish a course. For anyone else with a neuromuscular condition I would advise you to make sure you participate in activities which you find enjoyable; whether that be uni, or hobbies and passions such as sport.

For over 12 years I have participated in electric wheelchair sports, both at a local and national level. I compete in electric wheelchair soccer, hockey, and rugby. For a total of eight years I have attended and played at Nationals in Sydney, an achievement which I am very proud of. My greatest achievement at local level would be winning the 2015/16 Saturday League Best and Fairest, however I also hold the Minor Premierships I have won with team mates in high esteem.

For many years now I have been involved on the Western Electric Sporting Association (WESA) committee in a myriad of positions, ranging from Saturday League Coordinator, and in more recent years, as President. As a long time member of WESA I believe it is important to contribute back to an association which has provided wheelchair athletes so many opportunities.

Through sports I have made friendships which will last for life. Sports clubs are optimum environments for creating social networks, and in the case of someone like myself with DMD, it has been an invaluable opportunity to connect

with others with similar conditions. Through these social networks I have found it far easier to come to terms with my disability.

Additionally to playing sport I am also an avid spectator. I am interested in a large variety of sports, including AFL, soccer, cricket, tennis, basketball, and ice hockey. I have been a Fremantle Dockers member for 11 years, and a Perth Glory member for seven years.

I have also been fortunate in having the opportunity of travelling internationally. I first travelled to LA at the age of 10 courtesy of the fantastic Make A Wish Foundation to spend time at Disney Land in Anaheim. I have also travelled to Barcelona, San Sebastian, New York, Las Vegas, Paris, Lyon, Frankfurt, London, Wellington, Singapore, Milan, and Rome, and will be heading to Japan in a few months.

One of my most cherished moments was attending an El Clasico soccer match in 2012 between FC Barcelona and Real Madrid, at Camp Nou in Barcelona. The game was played at 10.30pm in front of over 95,000 people.

The great thing about Europe is the relative ease in which you can travel between countries by train. A majority of stations are equipped with chair lifts, that transport you from platform to train. I much preferred traveling by train, as you are able to stay in your chair, which is of course far more comfortable than having to sit in an unadjustable plane seat. Although plane flights may not be all that comfortable, I highly recommend international travel if you are able to.

One of my major passions in recent years has been music. My music tastes are fairly diverse, ranging from Indie, to Britpop, to Metal, to Hip Hop, to Hardcore, and everything in between. Since the age of 18 I have associated with the Straight Edge subculture; a lifestyle which involves refraining from alcohol and drug consumption. I have found being straight edge to be a largely positive experience, and one which stands against the inherent drinking culture in Australia. Along with an interest in music I have a strong

interest in food. My interest doesn't simply end at the consumption of food, but also the process of food creation, as well as the anthropology of food. Food in many ways is indicative of a nation's culture. Through food you can learn a great deal about how a country may consider food.

I personally am a pescetarian; someone who only eats poultry and fish meat. My sister is vegan, and being the primary home cook, I also eat a lot of vegan food. I have learnt to enjoy the vegan lifestyle. It is a common misconception that vegan food lacks flavour, as a vast majority of vegan food I have eaten has been packed full of taste. It is not the ingredients which influence the attractiveness of food, but the way in which it is cooked and prepared.

At home we have several pets; a German Shepherd named Hoss, a Norwegian Forest Cat named Brann, and two Fancy Rats named Tonyo and Pippin. I find it highly therapeutic interacting with the pets, who present us with nothing but compassion.

This year I am hoping to involve myself in volunteer work or an internship, preferably in refugee, Aboriginal, or social justice issues; areas I have tremendous interest in.

the condition

Duchenne Muscular Dystrophy (DMD) is the most common of the childhood neuromuscular conditions and affects approximately 1 in 5000 live male births worldwide. In rare cases DMD can occur in females (approximately 1 in 50 million). Symptoms usually appear between the ages of two and five, and include frequent falls, difficulty running or getting up off the floor, a waddling gait and enlarged calves.

DMD is caused by an insufficient level of a protein called dystrophin. This lack in dystrophin causes muscles to deteriorate and break down, leading to progressive challenges with muscle function and mobility.

While there is currently no cure, physiotherapy and medication (such as corticosteroids) can help control slow progression and improve quality of life. Many individuals who live with DMD lead full and happy lives.

The good news is that new gene-based therapies have recently emerged with noted advances in using conventional gene replacement strategies, RNA-based technology, and pharmacological approaches. In particular, antisense-mediated exon skipping has shown encouraging results and holds promise for the treatment of dystrophic muscle.

In September 2016, the U.S. Food and Drug Administration (The FDA) finally granted accelerated approval for Eteplirsen (Exondys 51) as a treatment for DMD. Many of you may be aware, this therapy has been a significant focus of the research undertaken by our Honorary Life Members Professors Steve Wilton and Sue Fletcher for the past 20 years.

While this is exciting step in the right direction for those with DMD who have a confirmed mutation of the dystrophin gene amenable to exon 51 skipping, it also provides significant hope for other DMD target exons and the development of future treatments across different genes and different conditions.

