Dear Committee

My son is a 14 year old who may not have long to live. He sleeps much of time as too tiring at school.We were told 3 months ago that the dilatation of all chambers of his heart is irreversible. The sizes of each of his atria and ventricles is 11/2 times size of those of man 165 kg but my son is 55 kg. I have been trying to tell cardiologists and family and school about my sons collapses for years but as intermittent and undiagnosed I was not believed and he was not investigated - (even though ambulance ECGs showed oxygen at p02 of 42-44 which is neardeath for 10 minutes)and now we have found he also has cardiomyopathy of unknown cause. He has been having bad collapses where his heart and breathing stop for prolonged times for 4 years. I love my son and he his popular at school and likes drawing and was good at football when he was allowed to play 5 years ago.

1. Long QT gene test

After I resuscitated him in early 2005 and due to some of his ECGs reports had 'borderline long QT' -
his blood was sent for gene testing for long QT by Womens and Childrens Hospital medical geneticist
'Long QT' is a condition where people of all ages can die suddenly and people with this condition
can be given a defibrillator pacemaker to save their life - so a quick diagnosis is useful.
Luckily the Womens and Childrens Hospital paid half of the $3000 for the gene test

- expense - this is expensive largely due as far as I know due to patent on test. It was much money to me as was 1/6 of my net income for the year and all i had in bank.

His blood was sent to New Zealand the closest palce that did the test -in 2005 but we have not had any result yet...which has been frustrating as he has been having bad collapses and we do not know cause.
Unfortunately the person who would have done the test in New Zealand died and had not trained anyone else. Then his blood was sent from New Zealand to a Scandanavian country possibly Denmark. I asked the Adelaide medical geneticist in 2006, 2007 and 2008 if he could email directly to the scandinavian country to ask how long it would take as my son was often dizzy but he did not as he said New Zealand was embarrassed about what had occurred and he did not want to email any of them and would not give me the email address. Then when I asked again about the progress of his blood test for gene testing in Scandinavia in mid 2008 I was told that Denmark were sorry that his blood was lost and never got tested. Another sample of my son's 2005 blood was sent from Adelaide to Melbourne in July 2008 for the gene testing of long QT that Melbourne became able to do. We have not heard any result. Luckily I did not have to pay twice. I have had to live with stress of not getting results of gene testing and not knowing if my son would be coming home from school each day.

Since then my son has had some very bad collapses and has had head injuries requiring CT scans of head and xrays of neck due to how badly he falls when his heart and breathing stop and has nearly died on a number of occasions . Due to lack of diagnosis since 2005 I have been ridiculed by people including cardiologists who have not seen his collapses and so have said due to him not being dead then it cannot be serious and did not investigate for years and now he has irreversible severe dilated cardiomyopathy. We have been left out of family gatherings and he and sister and myself have been left out of finances permanently on both sides of family as family did not believe there was anything wrong as no diagnosis. Three months ago a MRI of his heart showed gross enlargement of all chambers of his heart. It is not known what type of cardiomyopathy he has nor where it started as all parts of heart now dilated as advanced and it would have been useful to see which chamber dilated first. I was told today the cardiologist wants to talk to my son about his 'future' and is considering a biopsy of his heart but will not do a referral as i have requested weeks ago to pediatric cardiomyopathy centre on Melbourne or Sydney which can understand MRI heart and tissue doppler more than in Adelaide.

2. Marfans, loez-dietz, erlers-danlos gene testing

It was thought since 2005 that my son and my youngest daughter could have Marfans syndrome as my son was known to have a dilated proximal aorta since early 2005 - which was important as chance of rupture and my daughter had very flexible joints, big arm span and dislocated joints and fractures more than usual if fell over. I was told as my son did not have over 90% of marfans signs that I had to pay another gene testing of $3000 and I was told he should also have gene test for Loeys Dietz $800 and Erhlers Danlos was another $5000 for the basic part so total over $8000 and not all of erhlers-danlos testing. I did not have $1000 in bank as on disablity pension due my heart and pacemaker and cardiologist only lets me work up to 15 hr/ week due my severe arrhythmias and collapses. My son's cardiologist said his dilated aorta only mattered if due Marfans and to do gene test but I did not have the money. Another cardiologist said it does not matter what cause of dilated aorta but it was important as could rupture and may need operation. We were told if he has severe pain between shoulderblades it could be his aorta rupturing. Since then we were told the pain could be in chest of back or abdomen when aorta ruptures. I applied to Variety Club to pay some of the money for his Marfan/ erhlers -danlos/ Loeys-Dietz gene testing but after having application for some months they chose not to and did not even spell Marfan in any way like the word in rejection letter- which for some reason I found upsetting.

I was getting used to knowing the gene tests were there but not affordable due patents owned by companies. I was learning to give up in trying to find out what is wriong with my son due patents on gene testing mean not affordable

As there was no diagnosis the school would not believe he has a heart problem. The school will not keep a defibrillator at the school to save his life even though I bought one and it is meant to be able to be used even by an untrained 11 year old. I offered to pay St Johns to train teachers how to use it to but they will not have it at his school

As there is no definite full diagnosis of collapses and cause of dilated cardiomyopathy - the school refuse to allow an ambulance to be called from the classroom if he is not breathing and no heartrate or pulse and looking dead. A child has to find a receptionist who has done a first aid course (but told me she is no allowed to take a pulse) and then she looks at my son and then goes back to office and phones ambulance. Ambulance on last callout was told wrong information by receptionist so did not hurry. My son's teacher went home crying at lunch as he thought my son was dying and he was an experienced teacher who wrote 2 pages about my sons 'life-threatening collapse with no pulse nor breathing for prolonged time' and did not think he would survive until ambulance came 15 minutes later. My sons friends will not have him for a sleepover as parents too frightened he could die at their house

I have got used to knowing there is something that needs sorting out and serious with my sons heart and yet nothing is done. Earlier this year my youngest daughter was diagnosed clinically with Erhlers-danlos and I was advised it may be advisable for her not to have children. She is 15 and it will also impact on her work and relationships due to this genetic disease. She may have the type where arteries rupture anywhere in body. If she does get pregnant as well as children having the condition, her uterus could rupture. There is gene testing to see if it is the type that causes arteries to rupture or not but I do not need to do this as my son is most important how sick he is.
I have been told that there are gene tests for dilated cardiomyopathy as my son has, but not yet available commercially and i doubt he will be able to get in research as other times I reuested there were no free research gene tests for what was needed.I expect when gene testing for cause and type of dilated cardiomyopathy are available if he is alive, the gene test will be too expensive as these tests are patented so companies and shareholders become rich and the people whose lives depend on the tests cannot afford them.
This reminds me of CEOs of companies receiving million dollar bonuses or golden handshakes when others are retrenched
Why should the people who need to be able to have the gene tests be missing out? Why does our goverment allow this? I wanted my son to be able to grow up and what help has he had?

I have learned to give up on trying to get my son's heart problem sorted due to expensive gene testing we cannot afford and takes ages or is lost or too expensive or cardiologists saying as he is not dead, he does not need any tests and now it is looking like nothing can be done as so advanced cardiomyopathy too If he does have arrhythmogenic right heart dysplasia as cause for his dilated cardiomyopathy which I was told his cardiologist thinks is possible cause- then he needs a defibrillator pacemaker - there could be gene testing for this - but what is the point of seeing if there is as it will no doubt be too expensive? It is a pity there is no assistance with these expensive tests or even if there could be tax deduction but not only 1/5 of over $1500 Gene patenting - the people who get rich from this make me ashamed to be part of the same human race as them
It makes me realise the facade that our nation and other so called developed countries are civilised but instead consist of groups of greedy people each out for themselves - letting children and others die due their greed..

It is my view gene testing should not be allowed to be patented by companies - or if it must be - can only be cost to public of eg $100 and not many $1000s per person per test. It makes me feel nausea thinking of how much a profit and how little a life is worth for shareholdersand we are not talking treatment just investigation so can get a diagnosis so can see how to treat.

I feel I have let my son down even though I have been telling cardiologists that it is not faints he has and I know it is life - threatening and 'please investigate his heart' for 4 years. I did not get help for my son from gene testing due cost of patents or not yet available in Australia nor help from the cardiologists who said it could not be serious as my son was not already dead but that it was genetic.
Now they have said there is nothing they can do as his heart is so dilated.
I think the companies and people receiving profits from exorbitant gene patents should be identified.

My son is an innocent Australian child with hopes and dreams and a descendent of Alfred Deakin one of the Founders of Federstion of Asustralia and 3 times Primeminister. Why could not my son get the testing needed including gene testing before it was too late? Why is it likely he will never grow up to be an adult?

I could try to find where in world is gene testing for dilated cardiomyopathy but likely not available and would use what I have in bank now we do not have a house. If available and if affordable, the result - would it be back when my son is still alive ?

Dr Belinda Coyte