**In Response to Coordinated Care for Chromosome 22 abnormalities in Northern Ireland.**

There are currently over 80 diagnosed patients held on the Belfast City Hospital genetics register living in Northern Ireland. Conservative and dated studies show that 22q affects 1:4000 live births.

22q goes so underdiagnosed that our Northern Irish population of 1.8 million has a diagnostic rate of 1:22500. Putting that into perspective we should have a diagnostic register of over 450 people (according to conservative national statistics mentioned above)

Due to these figures 22q currently falls under the “Rare Disease” umbrella.

Newborn screening would identify 22q from the outset and would have a minimal price associated as the bloods already taken from 24,277 live births per year in N.I. (2013) could be used to screen for 22q.

Reproductive options such as Pre Implantation Genetic Diagnosis (PGD) could prevent future 22q births.

If a foetal heart condition is detected or a baby is born with a congenital heart defect they are generally routinely tested for 22q given the high prevalence of heart defects in those with 22q.

The lack of integrated coordinated care between public services is devastating. The lack of coordinated care within the health services has a huge and draining impact on the overall resources in Northern Ireland.

**Aims of this Business Case**

It is our intention to highlight the current need for coordinated care as well as provide a constructive solution which will be cost effective and ensure a better quality of life for those affected by chromosome 22 abnormalities and their families.

This business model will lay out the following three points.

1. The need to deliver coordinated care through a specialist clinic
2. Examples on how this “Work Smart” initiative will save the trust money and give those diagnosed a better quality of life
3. Costs involved within this business model

**1 The Need to Deliver Coordinated Care through a Specialist Clinic**

22q affects health and quality of life from birth through infancy and childhood to adult life with over 180 physical, functional and psychological associations. Care for patients is multidisciplinary and lifelong. Early recognition and optimised, integrated care can achieve much in the way of improving outcomes and supporting affected individuals and families.

In 2013 Max Appeal launched the “Consensus Document”. It is a comprehensive but practical and accessible information resource which has had contributions from Major Centres from across the UK stakeholder organisations, families and over 50 experts working in the major clinical fields associated with 22q. (Please find attached in ATTACHMENT 1 – CONSENSUS DOCUMENT)

The Consensus Document provides the base model for a coordinated care protocol in Appendix 2. Recommended Assessments. This lays out the assessments which need carried out - At Diagnosis – Infancy – Preschool – School Age – Adolescence – Adult.

An APPG has been established in 2014 in Westminster with the intention of making this consensus a clinical protocol within each trust.

There is a sufficient caseload of patients with chromosome 22 abnormalities (as provided in the forward) to build a recognised expertise. In fact it is surprising there is none already in place given the current diagnostic rate let alone the changes which are imminent with genetic diagnosis abilities.

There is currently no coordination of care from a 22q specialist or specialist centre in Northern Ireland for pre-birth, children, transitioning adults, adults or pregnant women.

**Pre birth**

There is no coordination of care with a specialist with 22q knowledge for the foetus development when a diagnosis is made or suspected. The commonality of this situation is inevitability likely to increase as those currently diagnosed with 22q conceive as inheriting 22q has 50/50 chance of transmitting the condition to offspring in any pregnancy.

**Infancy**

Upon diagnosis a paediatrician who may or may not have a specialised knowledge of 22q or know or use the Consensus Document will manage the care of each child. This means the level of care depends solely on a paediatrician’s awareness and more importantly, up-to-date knowledge of 22q. Onward referrals to services are not knowingly made to specialists with 22q knowledge.

**Children Transitioning into Adulthood**

This is an extremely delicate area and there is no specialised management of care or coordinated care by a specialist with 22q knowledge.

**Adulthood**

Upon diagnosis a GP is responsible for the coordinated care of an adult who may or may not have the ability to request onward referrals to appropriate services. If onward referrals are made they are not knowingly made to a specialist with 22q knowledge. When a diagnosis is made as an adult it falls on the individual to push for any type of test or assessment. It is often the case that these individuals may not be able to coordinate their own care in a manner required for a life-long condition.

**Pregnancy**

There is currently no available coordination of care with a specialist with 22q knowledge at this physical and emotionally vulnerable stage in a patient’s life.

**Personal stories which drive the need for coordinated care.**

Northern Irish 22q patient Rebecca Gleed

Rebecca has never had coordinated care, her symptoms of tremors, because of insufficient calcium absorption (common 22q problem) finally led to the diagnosis of Osteoporosis at 20 years of age. This diagnosis could have been prevented if the recommended calcium levels had been checked and Rebecca placed on the appropriate daily supplement dose of Calcium and Vitamin D.

Northern Irish 22q patient Rachael Wright

Rachel has never had coordinated care and her mother never knew about the high risk of mental health issues. (Patients with 22q have a 33% risk of developing severe mental health conditions such as psychosis). Her diagnosis of Schizophrenia in her early 20’s could have been treated in her early teenage years had this care already been established and the symptoms and concerns been highlighted to a specialist.

Northern Irish 22q patients Carol and Lewis Gibson

Carol was diagnosed in 2009 with 22q after her second child was born with a complicated heart condition then diagnosed with 22q. Since her diagnosis in 2009 Carol received no assessments whatsoever and no coordinated monitoring throughout her pregnancy. In November 2014 as her third child was being born Carol needed emergency surgery to save both her and her baby’s lives during childbirth, it is unclear yet as to the implications of the lack of coordinated care with this birth on Carol but what is clear is the delay in testing and diagnosing baby Lewis leading to hypocalcaemia and seizures a number of days after his birth.

Northern Irish 22q patient Lucia Cassidy has had reactive care with her parents raising concerns over and pushing for diagnosis on a number of syndromic conditions due to 22q. 22q was initially diagnosed at the age of 3 years 8 months. Almost 2 full years after a number of clinicians were involved with Lucia’s different medical needs. Non specialist 22q clinicians involved in her care have stated “a diagnosis is not as important as treating the symptoms”, highlighting the dangers of non-coordinated care.

**2 Examples on how this “Work Smart” initiative will save the trust money and give those diagnosed a better quality of life.**

Actual saving for the genetics department cannot be estimated as there is no current arrangement in place for a 22q clinic to compare with this proposed model. The overall cost saving can only be guessed as highlighted by the staggering examples below.

The impact on mental health that a 22 chromosome abnormality can have should be highlighted here, not just on the patient but the entire family due to the lack of coordination within the trust. A specialised clinic would be so aware of these needs and early intervention and information would be invaluable.

Specific examples

Example 1 - A 22q patient had an appointment on a Wednesday with her paediatric doctor to have her calcium levels checked, on the Friday she had another blood draw through a separate clinician to check her antibodies. These two separate blood draws required two separate appointment slots, made by two separate receptionists carried out by two separate clinical nurses sent by two separate couriers to the same lab for two separate results which could have been done on the appointment slot by the same nurse and carried by the same messenger where one set of results could be obtained. This is a common occurrence with all 22q patients. As well as the costs involved the highlight of the patients quality of life should be noted. Time off work/school was required, transport to and from the hospital, car parking fees, babysitting other children needed organised. With an appointment schedule of 3-5 hospital appointments PER WEEK for a 22q patient the coordination of appointments would be invaluable.

Example 2 – A 22q patient required a hip x-ray for an undetermined limp, she had a spine x-ray due to monitor the progress of the curve in her spine. These x-rays could have been carried out at the same appointment by the same radiologist and sent to the relevant clinician. All of the above mentioned costs and impacts to quality of life could have been worked smarter.

Example 3 – A 22q patient required video fluoroscopy to see how she swallowed her food, she had a second video fluoroscopy to check her palate. All of the above mentioned costs and impacts to quality of life could have been worked smarter had one single test been carried out and results issued to the separate clinicians.

Example 4 – A family with 3 22q patients all had 3 separate ENT appointments. Coordinated care could mean these three people could be seen on the same day reducing 3 separate appointments to 1 extended appointment. All of the above mentioned costs and impacts to quality of life could have been worked smarter.

To build a multidisciplinary team the same patients would be referred to the same interested clinicians who will then build on this specialist experience. By way of keeping up to date with relevant and recent research and articles and being a member of the professional body of the 22q Society no cost or additional training or would be required to build a specialist team.

Words from those directly affected by 22q;-

**Martina Connelly – Mother to Alesshandra (1) 22q patient**

*“A 22q clinic would mean someone I could turn to when I have a question about Alesshandra eg something she's doing that we think she shouldn't be or vice versa.*

*People could contact the care coordinator if they are worried about something instead of doctors/hospital being first port of call if they were a bit concerned (this would save the NHS money as it would relieve pressure on local services)”*

**Beverley Spence – Mother to Angus (14) 22q patient**

*“It would be a one stop shop with experts who would work together and learn from each other. Records would be based in one location*

*It would help raise the profile of 22q and over time help educate medical and social services professionals on the condition and help raise its profile.  
It would help families meet other families and all the benefits that this brings.  
It would ensure all the necessary tests are done after diagnosis and none missed out.  
Remembering back to when Angus was first diagnosed we had so many questions and no one to answer them as you know it’s still rare to meet a medical professional who has heard about the condition.   
With regard to saving money perhaps by having a clinic it would speed up diagnosis (of syndromic symptoms) and insure the right tests were done.”*

**Rebecca Gleed – 22q patient**

*“A 22q clinic would be great so the people working there already know about the syndrome.” – Often we as the patient know more about 22q than the Doctor and that can be frightening as they are supposed to be the ones caring for us.*

**Richard Henry – Father to Oliver 22q patient**

*“Concentrated checks and observations specific to the condition, with experts in the field.*

*This would save money as only a certain group of doctors would need to specialise in the condition - less time moving between doctors who possibly have to read and learn the symptoms and tested of the 22q family.”*

[**Kathryn Burns**](https://www.facebook.com/kathryn.burns.33865) **- 22q patient**

*“I think a 22q clinic is a great idea it would help a lot of people it would save a lot of money maybe more than you think I’m not sure how it would save money but I know it might thank you for asking me these questions”*

[**Zoe Carlile**](https://www.facebook.com/zoe.carlile) **Mother to Tom, 22q Patient**

*“A clinic would mean the world to me and my son it would make life and issues so much easier be able to share my problems with someone else and have support from somewhere I think it would save the NHS a fortune as is not a widely talked about syndrome and there is very little known about it, and if there was more awareness raised for 22q 11 doctors might find it easier to diagnose a lot quicker therefore saving the NHS a lot of money as they waste a lot of time with misdiagnosis!”*

***Gillian Cassidy* – Mother to Lucia, 22q patient**

*“A 22q clinic would bring a peace of mind that someone with a speaclist knowledge of 22q is looking after my daughter from infancy right through to adulthood, will know her case and history will have a sense of the impact 22q has on her and our family life. The idea of coordinated care would reduce the number of times we visit the hospital which will reduce the levels of stress put on our family and the need of social interventions such as the costs in providing respite care.*

*Being able to have advice on symptoms to look out for and information including the first signs of mental health problems as she reaches her teenage years would be a tremendous relief to her father, his deepest concern with this rare disease is that our daughter will end up needing institutionalised and given there is over 25% chance this could happen it is a very real fear.*

*Our daughter having 22q means that her individual symptoms need to be addressed with a specialist knowledge so that she is given the right care plan. Her care plan is different even though her symptoms may be similar to that of a typical person.”*

**3 Costs involved within this business model**

Commitment given from an established genetic clinic of \*1.5 days per calendar month including the receptionist and clinical nurse at an estimated cost of

**Cost of £25k per annum.**

Commitment given from one existing Lead Clinician to provide \*4 hours per week.

**Cost of £10k per annum**

Commitment given from one existing Genetic Counsellor to provide \*4 hours per week

**Cost of £5k per annum**

Commitment given from a Care Coordinator to provide \*20 hours per week

**Cost of £20k per annum**

Total yearly cost £60,000\*

\*Based on the current diagnosed rate of circa 80 patients

Roles and Responsibilities laid out in the attached Job Descriptions.

(Please find attached in ATTACHMENT 2)

**CONCLUSSION**

This Business case centres round the theme of coordinated care and a proposal on how to streamline and make the system work smarter. Through focusing on this theme we think it clearly incorporates and addresses the other 5 themes as laid out in the Consultation Document.

* To empower those affected by 22q through a recognised service of specialists dedicated to improving their care and quality of life
* The financial implications involved in identifying and preventing 22q under the current systems in Northern Ireland via new born screening and subsequently the overall plug in the drain of resources once 22q has been identified a care plan established including which by
* Its nature covers the coordination of care from before diagnosis including early intervention.
* The role of research as set out in the Regional Genetics Service draft service specification could include this business case as being a innovative cost saving model which could be used further afield throughout the UK and Ireland
* Close links have already been established between our Republic of Ireland counterparts who hope to also be able to implement this plan with the ability to work collaboratively where national forward referrals are required from both ends of the island.

If you require any further information on this proposal please contact me

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Attachment 1 – Consensus Document

<http://www.maxappeal.org.uk/knowledge/consensus_document>

**ATTACHMENT 2 – JOB DESCRIPTIONS OF CLINICAL TEAM**

**Lead Clinician**

* Identify a clinical slot in an established clinic
* Identify an established Genetic Counsellor
* Identify a Care Support Coordinator
* Identification of local clinical departments relevant to the coordination of care for those with a diagnosis, develop these when further suitable departments arise (if not already established within the local health board then identifying national relevant clinicians) \*national meaning ROI, England, Scotland and Wales
* Establish and maintain multidisciplinary network of local specialist clinicians to deliver effective care through high quality communication, coordination and planning skills.
* To ensure each patient receives the right treatment at the right time
* Ensure each patient is involved in the determination of an appropriate care plan with which to meet their needs.
* Will oversee referrals for the relevant diagnostic testing and assessments as laid out in the Max Appeal Consensus Document and in addition create a care plan for diagnosed pregnant females
* Will use the database of diagnostic tests carried out for each patient and results to coordinate care
* Will keep up to date on research/articles/conferences relevant to the diagnosis
* Will actively communicate with the Clinical Team
* Will already be established within the trust and have the coordination of care included within

their workload.

**Genetic Counsellor**

* Identify diagnosed patients via the register and ensure all newly diagnosed patients are included in the clinic register
* Organise and attend the monthly clinic where Lead Clinician will see all diagnosed patents in one day
* Inform patients of due Assessments, and refer them to the relevant specialist.
* Will create and maintain a database of diagnostic tests carried out for each patient
* Be a contact for the patients when they have a query regarding the clinic
* Will keep up to date on research/articles/conferences relevant to the diagnosis
* Will actively communicate with the Clinical Team
* Will already be established within the trust and have the coordination of care included within their workload

**Care Support Coordinator**

* Have access to the register and establish contact with current and newly diagnosed patients to provide information on all services available
* Will issue an advocacy and support declaration form which will confirm permission to act on their behalf both verbally and in writing
* Be able to provide patients with information on and help with;-
* hospital transport
* benefits
* housing
* education
* local, national and international charities and support groups
* Provide specific support within the care plan when a diagnosed female becomes pregnant
* Attend the monthly clinic and be a representative at the request of the patient at clinical appointments
* Will follow up on any questions or support required as a result of the clinic or further appointments
* Be available to patients via email, telephone and where required in person
* Will invite the multidisciplinary specialist clinicians to become members of the professional body of the 22q Society, record their applications, hold a copy of their certifications and remind them of renewals.
* Will source and share relevant research and articles to the patients and multidisciplinary team via a newsletter
* Will attend relevant conferences and pass on information to the families and professionals alike.
* Will actively communicate with the Clinical Team
* Will have experience of local 22q patients, have the ability to work from home, have the ability to travel, will commit to a contract with the trust.