Paper prepared for presentation to the Citizens Assembly at the request of Justice Mary Laffoy, dealing with a neonatologist's perspective of babies born with severe fetal anomalies.

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Introduction

Fetal abnormalities can range from the minor (pre auricular skin tag) to the severe e.g. anencephaly. Most are actively managed and often transferred for surgery to one of the paediatric centres shortly after delivery, some are initially managed in ICU in the Rotunda then care is redirected and some are offered purely comfort care and are managed on the ward with parents, some even making it home to meet siblings and extended family and friends. Some families with planning may opt to have their baby's heart valves harvested for transplant.

In my experience parents value time with their babies and something as simple as getting to bath their baby before they pass away can be very important. With some conditions, such as Patau's or anencephaly, it is clear the baby may not even survive pregnancy or labour, though some can survive hours and occasionally days after delivery and parents need to be prepared for this. With the advent of fetal anatomy scans between 20-24 weeks most complex fatal fetal anomalies are diagnosed well in advance of delivery and parents can be counselled jointly with us as neonatologists and the fetal medicine specialists.

On other occasions the prognosis is not so easy to predict and we may seek expert input from other paediatric specialists e.g. cardiology, MRI or neurosurgery, to guide the information we are giving families. Often one has to wait until the baby is born to assess how they respond to initial resuscitation, doing more detailed investigations after delivery to confirm the diagnosis and reassessing the actual baby can change the prognosis.

The advent of paediatric palliative care and neonatal nurses with a specialist interest in same, has improved how we manage babies with severe conditions and the experiences the families have at a very difficult time.

The Rotunda annual clinical report for 2015 will be available on the website and pages highlighted in appendix one should give a sense of the numbers of babies diagnosed each year in Rotunda with complex anomalies. The numbers transferred annually by the national neonatal transport are in appendix 2. I will now describe below four fictional cases outlining the types of scenarios we face as Neonatologists.

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**Cases described are fictional but the clinical scenarios would be representative of real-life patients.*

Case 1

Jane Kelly aged 37 presents with her first pregnancy. She has an uncomplicated first trimester. The anomaly scan at 22 weeks is suggestive of the baby having Edwards' syndrome. An amniocentesis performed 2 weeks later confirms same. The couple meets with one of the neonatologists following initial counselling with the fetal medicine team. Specific outcome and survival data are shared with the parents about the likely neonatal outcome of Edwards'syndrome. They are aware that this condition is incompatible with longer term survival and a clear care plan is put in place.

Baby David is born at 37 weeks weighing 2kg (4lb 6oz). He is nursed on the ward with parents where they have time for him to be baptised, meet his grandparents and have a bath. Specialist imprints of his foot and hand prints are taken and he is monitored by the midwives and paediatric team on call. He passes away 12 hours after delivery and the parents take him home to be buried. Bereavement follow up and counselling is arranged by the bereavement midwifery team but the parents decline same, as they feel all was well explained prior to the delivery.

Case 2:

Jane Kelly is a 28 year old lady expecting twins on her third pregnancy. At the 20 week anomaly scan it is noted that the second twin has a congenital heart defect that will require complex surgery. Jane's waters break at 27 weeks and she is admitted to hospital, given antenatal steroids and the consultant neonatologist comes to talk to her about the complications of prematurity and the added complication of twin 2 having a complex cardiac defect. The paediatric cardiologist is also informed of the admission with threatened preterm labour, as she was involved in doing the more detailed cardiac scans prior to delivery. The minimum weight a baby can undergo this complex surgery is 2.5kg (5lb 8oz). A plan is put in place to monitor the affected twin after delivery and see if medication can keep the in utero circulation going until the baby reaches a weight of at least 2kg (4lb 6.5oz).

The twins are born 36 hours later in good condition and are transferred to the NCIU. Twin 1 Mary weighs 1.1kg (2lb 6oz) and twin 2 John weighs 850g (1lb 13oz). John is immediately commenced on medication to keep his duct open (PDA) and high calorie intravenous nutrition (TPN) to maximise his growth. He has an echo within an hour of delivery by a neonatologist with a special interest in new-born echoes and is reviewed weekly by the consultant paediatric cardiologist who visits the Rotunda. Despite maximum care from 3 weeks post-delivery the PDA gradually starts to close. At this point he weighs 1.1kg (2lb 6oz) and is significantly below any weight where surgery is technically possible. Following detailed discussions with the parents, paediatric cardiology, consultant neonatologists and neonatal nursing staff all agree that to continue with intensive care would be futile.

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John is baptised, extended familyvisit and a memory box are made which includes photos and imprints of his hand and foot prints. He is transferred to a quieter section of the NICU. All medication and infusions have been stopped, he is taken off his CPAP machine and the parents get to hold him. Photos are taken without any equipment and the parents dress him in clothes they have chosen. He dies peacefully in his mum's arms 40 minutes later.

He is nursed in a cold cot overnight with mum and dad, before the parents take him home to be buried. They continue to visit and care for their surviving twin Mary, who is discharged home well some 5 weeks later weighing 1.8kg (3lb 15oz). They attend developmental follow up for Mary in the Rotunda with the same consultant who was the main carer for both twins. They are also offered formal bereavement follow up which they avail of to discuss in more detail the events leading up to John's passing away. Having all the information prior to his delivery, the continuity and consistency of care following his birth and the time, though brief, they had with him are all things they valued. They also appreciated though a "long shot" that he was given every chance to survive, without suffering or pain.

Case 3:

Jane Kelly is a 32 year old lady on her fourth pregnancy. She is booked at a centre that doesn't have fetal anomaly scanning available. She delivers baby Mary by spontaneous vaginal delivery at 39 weeks, weighing 3.5kg (7lb 11.4oz). The baby has difficulty breathing at birth and is noted to have multiple congenital anomalies. The baby is transferred to Rotunda by the neonatal transport team (NNTP), that night for immediate stabilisation and assessment. Mary is then transferred to PICU in one of the Children's hospitals for further assessment, as the cranial ultrasound is abnormal and the baby needs an MRI brain and tertiary specialists input from neurology, ENT for airway issues and genetics. Following over 2 weeks of intensive care it transpired that despite not having the typical facial appearance and low birth weight, Mary has Patau's syndrome and care is redirected. Mary dies peacefully in her father's arms some 20 minutes later.

Bereavement follow-up highlighted the difficulty the parents had in balancing the time they had with their daughter and the prolonged course of treatment and complex care she received until the diagnosis was confirmed. They expressed concern that they didn't have a detailed anatomy scan, which may have prompted amniocentesis and genetics results prior to delivery.

Case 4:

Jane Kelly is a 42 year old lady expecting her first baby following 5 rounds of IVF. She had non-invasive prenatal testing in the first trimester which was normal. There were some concerns raised from the anomaly scans concerning skull shape and amniocentesis was performed which revealed normal karyotype and microarray genetics results.

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Baby John was born at 38 weeks by elective section. He could not be intubated and so an alternative airway (LMA) was inserted. He was transferred to one of the Children's Hospital's for an emergency tracheostomy at which time fibre optics confirmed a laryngeal web. He had significant craniosynostosis and had multiple operations for same over the course of his first 12 months. While heart, lungs etc. were structurally normal he had significant abnormalities on his MRI brain and over the course of his first year it became apparent he had severe developmental delay, seizures and an inability to survive without intensive airway support. Following numerous multidisciplinary team meetings with the parents, John was taken home with the support of the palliative care team and spent three weeks at home before passing away peacefully. The parents had nursing support from the Jack and Jill Foundation as well as their local hospice.

During bereavement follow up the parents noted their frustration at a lack of a defined underlying genetic disorder, though they acknowledged advance testing was on-going in a laboratory in England. They appreciated all the multi-disciplinary input, the time they had with John and how the decision to redirect care wasn't made until we were certain of the prognosis.

Conclusion

I hope these cases highlight the range and complexity of fetal anomalies we deal with in tertiary NICUs in Ireland. I hope they highlight how anatomy scanning and combined MDT meetings, as well as prenatal testing can help guide and prepare parents, although not always conclusively. Mostly, I hope the appendices that will be made available on the website to accompany this paper highlight that most babies born in Ireland with anomalies survive and that with the appropriate trained bereavement and palliative care teams, those who don't can be managed in a compassionate and caring way.

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Glossary

CPAP	Continuous Positive Airway Pressure
CVS	Chorionic Villous Sampling
ENT	Ear Nose and Throat
ELSCS	Elective Section
EmLSCS	Emergency Section
IVF	In Vitro Fertilisation
LMA	Laryngeal Mask Airway
MDT	Multidisciplinary Team
MRI	Magnetic Resonance Imaging
NICU	Neonatal Intensive Care Unit
NNTP	National Neonatal Transport Programme
PDA	Patent Ductus Arteriosus
PICU	Paediatric Intensive Care Unit
SVD	Spontaneous vaginal Delivery
TPN	Total Parental Nutrition
Trisomy 13	Patau's Syndrome
Trisomy 18	Edward's Syndrome
Trisomy 21	Down's Syndrome

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Appendix 1

Tables from Fetal medicine {P.141) and Paediatric {P. 69), departmental reports from Rotunda Annual* Clinical *Report 2015 {PDF on website)

Major Fetal Structural Abnormality diagnosed in Rotunda 2015:

Excluding soft markers and chromosomal abnormalities, 209 cases of major structural abnormalities were detected and followed.

Abnormalities detected based on RCOG/RCR Classification

Central Nervous System (CNS) (excluding choroid plexus cyst)	32
Head & Neck (including hygromata)	27
Cardiovascular system (excluding echogenic foci and untreated arrhythmias)	43
Renal (excluding pelvic dilatation of<10mm)	48
Abdominal contents (Including anterior abdominal wall defects and excluding echogenic bowel)	17
Skeletal	24
Thoracic (excluding cardiac abnormalities)	16
Others	2
Total	**209**

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Prenatal Diagnosis Clinic

In 2015 1,290 new patients attended for Prenatal Diagnosis. There were 3,089 attendances as some patients were followed longitudinally. All patients had an ultrasound scan. In addition the following tests were performed:

Combined First Trimester Screening	416
Non Invasive Prenatal Testing (Cell Free Fetal DNA)	651
Amniocentesis	114
Chorionic Villus Sampling (CVS)	80

Of the 194 diagnostic procedures performed, there were 63 abnormal results representing 32.5% of invasive tests.

Abnormality	**CVS**	**Amnio**	**Total**
Trisomy 21	13	7	20
Trisomy 18	8	8	16
Trisomy 13	0	3	3
45X	3	1	4
Triploidy	2	2	4
Mosaic	2	3	5
Translocation	0	1	1
Deletion	0	1	1
Di George	0	1	1
Sickle Cell	1	0	1
Klienfelter	0	1	1
Pallister Killian	0	1	1
Inversion	0	1	1

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SMA	1	0	1
Cystic Fibrosis	1	0	1
RYR1 Mutation	0	1	1
Smith Lemi Opitz	1	0	1
Total	**32**	**31**	**63**
Failed Culture	2	0	2

Note: Six patients with positive NIPT results declined invasive testing but the following results were confirmed postnatally:

Trisomy 21	4
Klienfelters	1
Trisomy X	1

There was 1 false positive for T13 on NIPT which was a normal result on CVS .

Main Indications for Admission to the Neonatal Unit 2015 (n=1,311)

RESPIRATORY SYMPTOMATOLOGY	523
PREMATURITY < 37 WEEKS	357
JAUNDICE	360
LOW BIRTH WEIGHT < 2.5Kg	446
HYPOGLYCAEMIA	241
CONGENITAL ABNORMALITIES	**240**
SUSPECTED SEPSIS	40
NEONATAL ABSTINENCE SYNDROME	28
SEIZURES	8
HIE	29
GASTRO-INTESTINAL SYMPTOMS	15
SOCIAL	5
DEHYDRATION	16

*Some Infants are assigned more than one reason for admission

Neonatal Mortality Rotunda 2015 congenital anomalies (n=18)

Birth Wt	Gestation	Delivery	Age	Diagnosis
2.34kg	40+0	SVD	4 days	Trisomy 13, Holoprosencephaly
2.77kg	38+6	SVD	2 days	Turner's Syndrome, Hypoplastic left heart
2.42kg	40+6	LSCS	<1 day	Osteo Imperfecta
2.0kg	31+0	EmLSCS	<1 day	Large omphalocoele, cord avulsion, multiple contractures
1.9kg	37+0	SVD breech	<1 day	Renal agenesis
2.49kg	38+4	ELSCS	<1 day	Right congenital diaphragmatic hernia, pulmonary hypoplasia
3.48kg	39+6	SVD	<1 day	Pulmonary hypoplasia, severe skeletal anomalies,
1.36kg	34+0	SVD	<1 day	Bladder outlet obstruction, Potter's Syndrome
3.38kg	38+5	forceps	3 days	Diaphragmatic eventeration, pulmonary hypoplasia
1.75kg	40+0	SVD	<1 day	Lumbosacral myelomeningocoele, renal agenesis
2.54kg	35+6	SVD	<1 day	Thanthorphic dwarfism
2.87kg	42+2	ELSCS	1 day	Anencephaly
1.1kg	31+4	SVD	<1 day	Congenital diaphragmatic hernia, Cornelia de Lange
1.07kg	32+0	SVD	<1 day	Trisomy 18
1.83kg	41+1	SVD	<1 day	Trisomy 18

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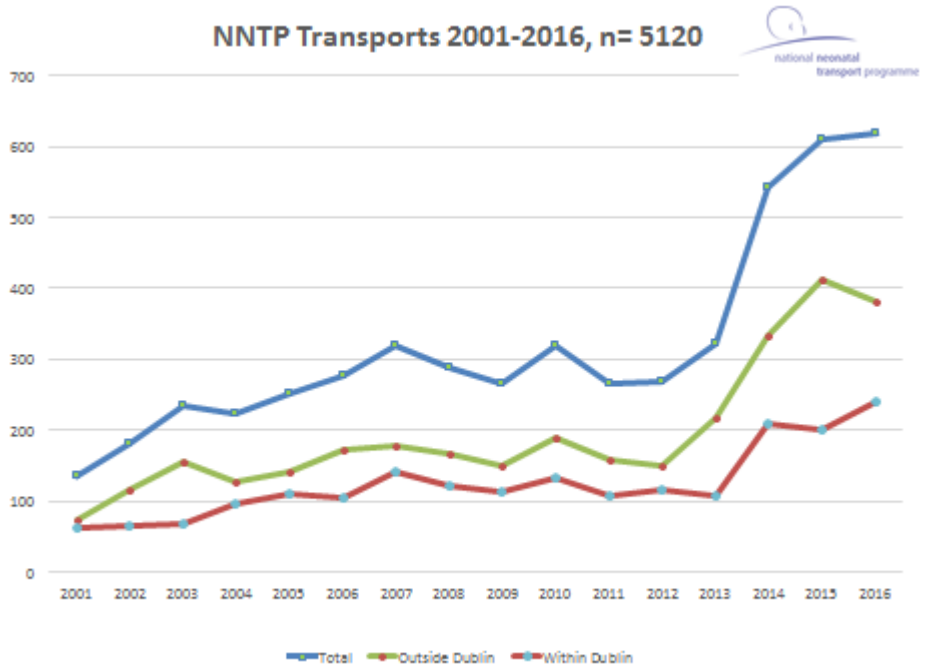
Appendix 2

National data of the numbers of babies transferred annually with congenital anomalies.

NNTP Transports 2014 & 2015
Primary Clinical Reason for transfer

national neonatal transport programme

	Medical	Surgical	Cardiac	Neurological
2014	237	130	124	52
2015	289	145	133	44
%	45.6%	23.8%	22.3%	8.3%



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