Who We Are

SOFT UK was founded in 1991 as an independent charity to provide support and information to families affected by Trisomy 13 (Patau's), Trisomy 18 (Edwards' Syndrome) and their related disorders.

Information & Support for Families:

- Through prenatal diagnosis
- During pregnancy
- Before, during and after a termination
- Following a miscarriage
- Caring for a baby, child or adult with Trisomy 13/18
- Through loss, bereavement and grief
- When considering future pregnancies

Information & Support for Professionals:

- Signposting to the most recent research and information on Trisomy conditions
- Access training and information opportunities for professionals
- Share learning with other professionals

SOFT UK Professional Advisors

Dr Nora Shannon Consultant Clinical Geneticist, Nottingham City Hospital
Ali Brett Bereavement Midwife, Ipswich Hospital
Dr Una MacFadyen Retired Consultant Paediatrian, Forth Valley NHS
Joan Morris Professor of Medical Statistics, St George's, University of London
Dr Lucy Kean Consultant Obstetrician, Nottingham University Hospitals
Erica Brown Vice President, Acorns Children's Hospices, West Midlands. Principal Research Fellow, Centre for Children and Families, University of Worcester, Trustee The Myriad Centre, Worcester
Professor John Carey Geneticist & Paediatrician. Founder & medical advisor of SOFT USA
Barbara Rosenthal MBACP Counsellor in private practice. Trainer for Cruse Bereavement Care

Lived Experience of Trisomy

Every aspect of SOFT UK's work is grounded in the Lived Experience of our families.

"Phoebe smiles, laughs, has started to baby talk since having her hearing aid, moves her arms and hands well, starting to move her legs, loves her baby music classes we go to, loves being cuddled, finds it hilarious getting changed, enjoys baths and most of all having raspberries blown on her forehead." Hollie, Mum to Phoebe Amber, Full T18, Age 1

"Our lives are better, richer and fuller with Isabel in them. She is full of love and joy, and absolutely living life to the full. Yes, she has significant developmental delays and is certainly medically fragile, but hers is very much a life worth living." Alison, Mum to Isabel, Full T18, age 8.

"This week at (4yrs, 3mths & 23 days) he has taken his first steps independently. (without his walker or our assistance.) The determination our kiddies have is incredible." Alison, mum to Ewan, Full T13, age 4.

References

1. National Congenital Anomaly and Rare Disease Registration Service, Congenital Anomaly Statistics, 2017
2. Cavadino A & Morris JK (2017) Am J Med Genet, 174: 953-958
3. Wu J, Springett A., Morris JK (2013) Am J Med Genet A. 2013 Oct;161A(10):2512-8 Meyer RE et al. (2016) Am J Med Genet, 170A: 825-837
4. Meyer RE et al. (2016) Am J Med Genet, 170A: 825-837
5. Nelson KE, Rosella LC, Mahant S, Guttmann A. (2016) J Am Med Assoc, 316: 420-428
6. Pont SJ et al. (2006) Am J Med Genet, 140A: 1749-1756
7. Patterson J et al. (2017) Am J Perinatology, 34(09):887-894
8. De Souza E et al. (2009) Am J Med Genet, 149A:29716-2722 Donovan JH, Krigbaum G, Bruns DA (2016) Am J Med Genet, 172: 272-278

SOFT UK
Support Organisation for Trisomy 13/18

Information Leaflet for Professionals

Edwards' Syndrome(Trisomy 18) & Patau's Syndrome (Trisomy 13)

SOFT UK is a registered charity
No. 1002918 (England & Wales) SC043341 (Scotland)
Registered address: 48 Froggatts Ride, Walmley, Sutton Coldfield, B76 2TQ

Email: **enquiries@soft.org.uk**
Web: **www.soft.org.uk**

Prevalence

Reporting on prevalence of Trisomy in the UK has been inconsistent since 2013 when the NDSCR closed. NCARDRS is due to publish England wide data by 2020/21 with CARIS in Wales and CARDRISS in Scotland following thereafter. The NCARDRS report states: "The prevalence based on national data is likely overestimated, as there is a higher proportion of babies where we do not know the gestation and outcome of the pregnancy in new NCARDRS regions compared to the 7 established regions."

Between 1 January 2015 to 31 December 2017 inclusive, there were 1,668 babies with Edwards' syndrome and 690 babies with Patau's syndrome delivered in the whole of England.

Studies indicate that prevalence increases with maternal age.

Trisomy diagnoses in pregnancy: outcome data for seven NCARDRS reporting regions 2015-2017 [1]

Antenatal Diagnosis | Postnatal Diagnosis

	Trisomy 13		Trisomy 18	
	Number	%	Number	%
Total diagnoses	277		697	
Total Antenatal diagnoses	178	64.3	503	72
TOPFA	161	90.4	437	86.9
Miscarriage (20 - 23wk)	1	0.6	6	1.2
Stillbirth (24wk+)	8	4.5	25	5
Live Birth	4	2.2	19	3.8
Unknown	4	2.2	16	3.2
Total postnatal diagnoses	99	35.7	194	27.8
TOPFA	58	58.6	108	55.7
Miscarriage (20 - 23wk)	18	18.2	16	8.2
Stillbirth (24wk+)	3	3	14	7.2
Live Birth	20	20.2	56	28.9
Unknown	0	0	0	0

Full, Partial & Mosaic

The symptoms of trisomy occur on a spectrum. Although children with trisomy share common characteristics, each child is unique.

Symptoms and the severity of symptoms will depend upon whether the child has full or mosaic trisomy, the number of cells affected by the trisomy as well as whether the trisomy is a partial or due to a translocation.

Screening does not indicate whether a child has full/ mosaic/partial forms of Trisomy. It is impossible to tell from screening what the full spectrum of physical, learning and developmental disability will be for each child. A pre or post natal diagnostic test will give more information from this point of view.

Full Trisomy
The extra chromosome is in every cell in the baby's body.
Mosaic Trisomy
The extra chromosome is only in some of the baby's cells.
Partial/Translocation

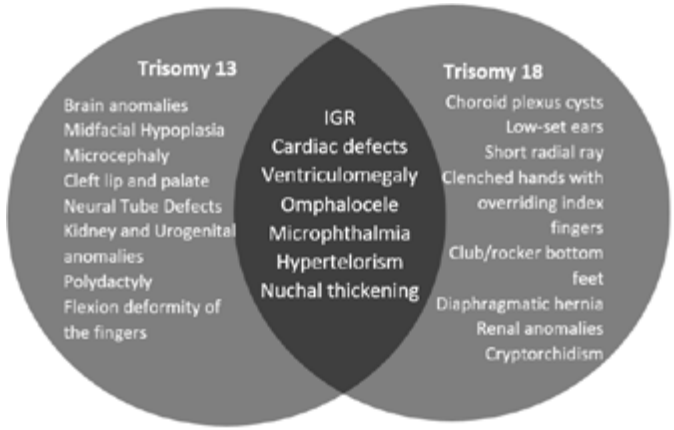
Median Survival

A high proportion of pregnancies diagnosed with trisomy 13 and 18 will end in miscarriage. [2] Median survival times are 12.5 days for trisomy 13 and 9 days for trisomy 18. [3]

Age	Wu *et al.*, 2013 (N=309) % surviving	Meyer *et al.*, 2016 (N = 1,113) % surviving
1 week	60	52.5
1 month	39	37.2
3 months	20	NR
1 year	**8**	**13.4**
5 years	NR	12.3

"Survival among children with T13/18 is higher than those previously reported [4]*, consistent with recent studies reporting improved survival following more aggressive medical intervention for these children* [5]*."*

Common Major Structural Malformations [6]



Neonatal Management

Parents who decide to continue with a pregnancy should have an opportunity to discuss their baby's diagnosis and plans for delivery and aftercare. Common issues after delivery include feeding difficulties, gastro oesophageal reflux, apnoea and problems linked to congenital anomalies. The scope of interventions being offered is increasing, and the spectrum of care for infants with trisomy 13 and 18 illustrates the need for individualized counselling that is on going, goal directed, collaborative and responsive. [7]
There are emerging studies that suggest improved outcomes when affected infants and children are given necessary medical interventions. [9]

Recurrence Risk

There is a small increased risk of the same trisomy after a previous pregnancy with trisomy 13 or 18. There may also be a small increased risk of trisomy 13 or 18 for women with a previous pregnancy with trisomy 21. [8]

The option of a prenatal diagnosis in subsequent pregnancies should be discussed.