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SOFT UK is a registered charity
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Who We Are

About Us

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.

We provide information and support to 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

Why contact SOFT UK?

- Build links with professionals who have a specialist interest in these and their related conditions
- Find affected families who are willing to be contacted by other families to share experiences
- Access current research on trisomies 13 and 18
- Become more involved with the work of the charity in supporting families

SOFT UK advisors

Dr Nora Shannon Consultant Clinical Geneticist, Nottingham City Hospital
Dr Una MacFadyen Consultant Paediatrician & Neonatologist, Stirling Hospital
Professor Joan Morris Director, National Down Syndrome Cytogenetic Register, Wolfson Institute of Preventative Medicine
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Professor John Carey Geneticist & Paediatrician. Founder & medical advisor of SOFT USA
Barbara Rosenthal MBACP Counsellor in private practice. Trainer for Cruse Bereavement Care

SOFT UK
Support Organisation for Trisomy 13/18

Information Leaflet

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

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Prevalence

Whilst Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau's syndrome) are very rare conditions, they are the second and third most common autosomal abnormalities after Trisomy 21 (Down Syndrome).

In England and Wales n 2013, the rate was 0.3 per 1000 births for Patau Syndrome and 0.7 per 1000 births for Edwards' Syndrome. Only 180 diagnoses of Trisomy 13, and 474 diagnoses of Trisomy 18 were made in 2013, resulting in 19 live births of children affected by Trisomy 13, and 40 children affected by Trisomy 18. [1]

Studies indicate that prevalence increases with maternal age.

Figure 1. Prevalence of T13, T18 and T21 according to maternal age

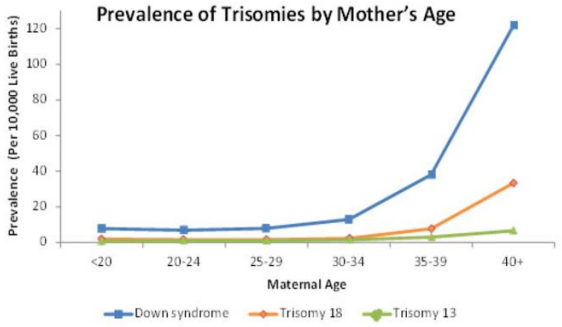


Table 1. Number and % of diagnosed cases of T13 and T18 in England and Wales in 2013

		Trisomy 13		Trisomy 18	
		No.	%	No.	%
Prenatal	Termination	130	72	358	76
	Live birth	4	2	11	2
	Still birth/Miscarriage	10	6	17	4
	Unknown	19	11	55	12
Postnatal	Live birth	14	8	27	6
	Still birth/Miscarriage	3	2	6	1

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 translocational trisomy.

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.

Full Trisomy - The extra chromosome is in every cell in the baby's body.
Mosaic Trisomy - The extra chromosome 18 is only in some of the baby's cells.
Partial/Translocation - The child has only part of an extra chromosome.

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

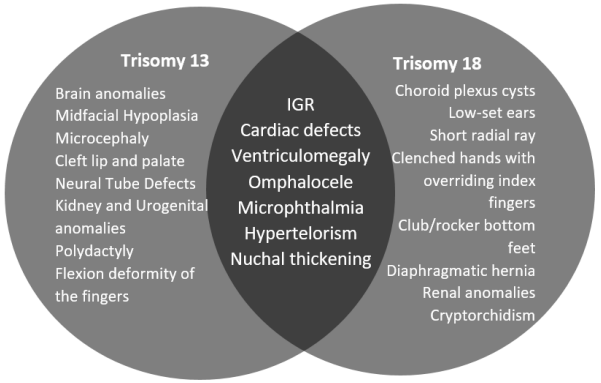
"Survival among children with T13/18 is higher than those previously reported, consistent with recent studies reporting improved survival following more aggressive medical intervention for these children."

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. [6]

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

Common Major Structural Malformations



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.[8]

There are emerging studies that suggest improved outcomes when affected infants and children are given necessary medical interventions.[9]