



**PORTSMOUTH
DOWN SYNDROME
ASSOCIATION**

Delivering a **DIAGNOSIS** OF DOWN SYNDROME



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A guide for healthcare professionals

Down syndrome is the most commonly occurring chromosomal condition, present in 1 in 800 babies born each year. Historically, the majority of Down syndrome diagnoses have been made postnatally, but due to recent advancements in prenatal screening and the introduction of the non-invasive pre-natal test (NIPT), an increasing number of diagnoses are now made prenatally.

Physicians delivering a diagnosis of Down syndrome have the opportunity to shape the early phase of a family's journey to understanding the diagnosis, but in a survey of families who had received a diagnosis, 24% reported negative experiences which included:

- Lack of empathy when explaining the diagnosis (70%)
- Pressure to have had prenatal testing (15%)
- Lack of information provided, and physician's lack of knowledge of Down syndrome (11%)
- Lack of support and low expectations for their child (11%)

Studies have shown that nearly all mothers reported feelings of initial shock, anger and fear following a diagnosis. Yet, these same mothers indicated that if physicians were to implement a few simple measures, as research suggests, the experience could be more sensitive to their emotions and needs. This research has shown that these key points should be considered when delivering a diagnosis of Down syndrome:

Postnatal Diagnosis

- Parents prefer to hear the news from the most knowledgeable professional(s); usually the physician
- Have the conversation in a private setting as soon as physician suspects a diagnosis of Down syndrome
- Deliver the news to parents together when possible, in a sensitive manner
- The infant should be present if appropriate and referred to by name
- Use person-centric language, 'a baby has Down syndrome' rather than a 'Downs baby' or 'Down syndrome child'. Avoid referring to the condition or individual as 'Downs' (see terminology sheet)
- Begin the conversation with positive words, such as "Congratulations on the birth" of the child
- Avoid using negative phrases language such as, "I'm sorry" or "Unfortunately I have bad news"
- Provide accurate and up-to-date information, including opportunities to connect with other families and support agencies
- Limit discussions to medical conditions that the infant has or might develop within one year
- Follow-up appointments should be arranged, as desired and needed



Prenatal Diagnosis

- Results from the prenatal screening should be explained in neutral language. Avoid negative language such as 'risk of Down syndrome' or 'positive' or 'negative' result
- Prior to screening, discuss all reasons for prenatal diagnosis
- Healthcare professional(s) most knowledgeable about Down syndrome should deliver the news in person if possible
- If an in-person visit is not possible, the news should be delivered over the phone at a pre-arranged time
- Avoid using negative phrases language such as, "I'm sorry" or "Unfortunately I have bad news"
- Use person-centric language, 'foetus with Down syndrome' rather than a 'Downs foetus' or 'Down syndrome baby'. Avoid referring to the condition or individual as 'Downs' ([see terminology sheet](#))
- Discuss all options available to parents, including continuing the pregnancy
- Give balanced, accurate and up to date information including opportunities to connect with support agencies, and be able to answer questions: What is Down syndrome? What causes the condition? What are realistic expectations for individuals with Down syndrome today?
- Offer up-to-date materials or bibliography
Down Syndrome Association UK:
www.downs-syndrome.org.uk/for-new-parents/being-pregnant/
- Make follow-up appointments, including specialists, as needed

Outcomes

The news of a diagnosis of Down syndrome, may trigger an emotional response from families. It is important to allow time for silence and time to express these emotions and offer the family time alone.

Likewise, research has shown that when families have time to bond with their child and get to know them as they grow and develop, those possible feelings of fear or worry, evolve into love and pride.

According to a survey conducted of more than 3,000 parents:

- **99%** love their child with Down syndrome
- **97%** are proud of their child with Down syndrome
- **79%** felt their outlook on life was more positive because of their child
- **94%** of siblings love their brother/sister with Down syndrome and are proud of them
- **4%** regretted keeping a child with Down syndrome





Alice's Story

When Edward was diagnosed we were in a private room, and my husband and I were together holding Edward. The consultant was kind, factual and gave us the medical diagnosis straight away. He was supportive and gave us time to take this in before offering further information.

Jo's Story

We had a prenatal diagnosis and our experience would have been a much more positive one if first person language and positive terminology had been used. Our children are not a diagnosis or just Down syndrome, they are sons and daughters, brothers and sisters, grandchildren, friends. My son was truly loved before our diagnosis; he is Louis.



Ken's Story

The geneticist used outdated terminology from the start. He told us how our son would never be independent and emphasised how little he would be able to achieve. He then went on to list every serious medical condition that our son might encounter throughout life.

Rachael's Story

Max's paediatrician was great. She shared her own positive experience with us of being a carer for a family member and put us in touch with our local support group, whose early advice was invaluable.

