

Student Number: 2405438

Word count : 1632

Introduction:

Williams–Beuren syndrome is a rare neurodevelopmental condition caused by a microdeletion on chromosome 7q11.23, which disrupts the elastin gene and leads to a distinctive combination of physical, cognitive, and behavioural features (Ewart et al., 1993; Pober, 2010). Because William syndrome affects multiple areas of development, from cardiovascular health to learning and social behaviour, it is often recognised through its unique profile rather than a single defining symptom (Mervis and John, 2010; Axelsson et al., 2013). This poster aims to build clearer awareness of what Williams–Beuren syndrome is, how it presents, and why early recognition is so important. By bringing together reliable research and highlighting the key challenges and strengths associated with the condition, the intention is to support a better understanding among those working in health, education, and social care. Ultimately, the goal is to ensure that individuals with Williams–Beuren syndrome are identified early and receive the specialist support that can make a meaningful difference throughout their lives (American Academy of Pediatrics, 2001).

Barriers:

Individuals with Williams Beuren syndrome often experience significant barriers that affect their development, learning and overall quality of life. One major barrier relates to the high prevalence of cardiovascular complications, which occur in approximately 70 to 80 percent of individuals and frequently require ongoing medical monitoring or surgical intervention (Collins, 2018; Pober, 2010). These health demands can disrupt daily routines, limit participation in physical activities and place additional pressure on families who must manage complex medical appointments. A second barrier involves the distinctive cognitive and behavioural profile associated with William syndrome. Although individuals often show strengths in verbal communication and sociability, they commonly experience difficulties with visuospatial tasks, attention and anxiety, which can make educational environments challenging (Martens et al., 2008; Mervis and John, 2010). Furthermore, hypersociability, reported in up to 90 per cent of cases, can increase vulnerability in social situations as individuals may struggle to recognise social cues or potential risks (Järvinen et al., 2013). Limited access to specialist services such as speech therapy, occupational therapy and psychological support can further widen developmental gaps, particularly in areas where funding or provision is inconsistent (Fisher and Morin, 2017; van Herwegen and Ruby, 2020). Together, these barriers highlight the need for coordinated, long term support across health, education and social care to ensure that individuals with Williams Beuren syndrome receive appropriate and timely intervention.

Williams-Beuren Syndrome

Supporting strategies:

Children with Williams Beuren syndrome benefit from early, targeted support that builds on their sociability and enthusiasm for learning while addressing areas where they may face challenges. Mervis and Velleman (2011) note that communication can develop unevenly, so early language intervention, visual supports and predictable routines can help children stay engaged. Sensitivity to sound and difficulties with changes in routine are also common, and Jones et al. (2000) explain that these can affect concentration and participation in busy early years environments. Although sociability is often a strength, Davies, Udwin and Howlin (1998) highlight that children may struggle with social boundaries, so modelling interactions and using social stories can support safer peer relationships. Motor development can also be an area of weakness, and Hocking et al. (2015) suggest that physiotherapy, occupational therapy and hands-on activities can help strengthen coordination and independence. A further challenge is the lack of consistent funding for specialist support. Fisher and Morin (2017) point out that limited resources can make it harder for settings to access speech and language therapy, one-to-one support or training for staff. This can affect how well learning strategies are implemented, meaning some children may not receive the level of support they need. Strong communication between home and early years settings helps maintain continuity, but adequate funding remains essential to ensure children receive the specialist interventions that make the biggest difference.

Statistics:

Williams–Beuren syndrome is a rare genetic condition, affecting around 1 in 7,500–10,000 people (Pober, 2010; Morris, 2017). Research shows that 70–80% of individuals develop heart problems, with supravalvular aortic stenosis being the most common and often the earliest recognised feature (Collins, 2018). In addition, around 75% of people have some level of intellectual disability, usually within the mild to moderate range, which can influence learning, problem-solving and independence (Martens et al., 2008). One of the most distinctive characteristics of the syndrome is the social behaviour often seen, as up to 90% of individuals are described as extremely friendly, talkative and emotionally sensitive, a pattern Järvinen et al. (2013) link to differences in social-emotional processing. Physical health concerns are also common; for example, 15–45% of children experience episodes of high calcium levels, particularly in infancy, which can lead to feeding difficulties and irritability (Morris, 2017). Sleep problems, anxiety and sensory sensitivities are also frequently reported, adding further complexity to daily routines and learning experiences (Ng et al., 2021). Altogether, these statistics highlight how wide-ranging the condition is and demonstrate why people with Williams–Beuren syndrome often require support from a variety of health professionals, including cardiologists, speech and language therapists, occupational therapists and educational specialists.

Diagnosis:

Diagnosing Williams Beuren syndrome usually happens after birth because current prenatal screening cannot reliably detect the specific microdeletion on chromosome 7q11.23. Strømme, Bjørnstad and Ramstad (2002) explain that most children are identified only when early developmental differences begin to appear, which highlights a significant limitation in early detection. These may include feeding difficulties, delayed motor milestones or cardiovascular problems such as supravalvular aortic stenosis, which Del Pasqua et al. (2009) describe as one of the most common early indicators. However, these early signs are not unique to Williams Beuren syndrome and can overlap with other developmental conditions, making early recognition challenging for practitioners. Early behavioural traits can also be noticeable, and Jones et al. (2000) note that children often show an unusually sociable manner and heightened sensitivity to sound, which can influence their early learning experiences and interactions. Yet relying on behavioural traits alone is problematic, as sociability can be misinterpreted as a strength rather than a potential diagnostic clue, leading to delays in referral. A formal diagnosis requires genetic testing, and according to Schubert (2009), methods such as chromosomal microarray analysis and multiplex ligation dependent probe amplification are now widely used because they accurately detect the 7q11.23 microdeletion. Although Morris (2017) adds that fluorescence in situ hybridisation may still be used in some clinical settings, this method is increasingly viewed as less sensitive than newer technologies. A critical issue is that access to advanced genetic testing varies between regions, meaning some families may experience delays or inconsistent diagnostic pathways. Early diagnosis is especially important in the early years because it helps families understand their child's developmental profile and ensures that appropriate medical, educational and social support can be put in place to reduce the impact on learning and development, as highlighted by Martens, Wilson and Reutens (2008). However, the effectiveness of early intervention is often limited by funding, availability of specialists and the level of awareness among early years practitioners. This means that even when a diagnosis is made, the support offered may not always meet the child's needs.

This image provides an overview of Williams Beuren syndrome by illustrating the key signs and symptoms that may be noticed in children. It highlights features such as:

- distinctive facial characteristics
- heart conditions
- feeding and digestive difficulties
- joint and mobility issues
- sleep disturbances
- raised calcium levels
- Severe anxiety
- heightened sensitivity to sound

Overall, the poster shows how the condition affects multiple areas of development and emphasises the importance of early recognition and coordinated support.



Lucy Pitt (2026)

conclusion:

Overall, the information presented shows that Williams Beuren syndrome is a complex genetic condition that affects many different areas of development, health and behaviour. The syndrome is rare, yet the range of physical, cognitive and social characteristics makes early identification essential for ensuring that individuals receive the right support. The high rates of cardiovascular problems, intellectual disability and metabolic difficulties highlight the need for regular medical monitoring, while the distinctive social and behavioural profile demonstrates why educational and psychological support are equally important. Barriers such as limited access to specialist services and the challenges linked to anxiety, attention and hypersociability further emphasise the importance of coordinated, long term intervention. By increasing awareness and understanding of Williams Beuren syndrome, professionals across health, education and social care can work together more effectively to improve outcomes and support individuals and their families throughout their lives.

Please find attached the image consent form for sharing a poster containing an image of Oliver Pitt (my brother). Consent has been provided and signed on behalf of myself (Lucy Pitt), my mother (Suzanne Pitt), and my father (Jonathan Pitt).

I can confirm that we give
Lucy Pitt consent to use the
photograph of our son Oliver for
her assignment.

Suzanne Pitt
S Pitt

Lucy Pitt
LPitt

JKP
JONATHAN PITT

Reference list:

American Academy of Pediatrics (2001) 'Health care supervision for children with Williams syndrome', *Pediatrics*, 107(5), pp. 1192–1204. [Online] Available at: <https://publications.aap.org/pediatrics/article/107/5/1192/66694/Health-Care-Supervision-for-Children-With-Williams> (Accessed: 4 May 2026).

Axelsson, S., Kjaergaard, S., Heiberg, A. and Boman, U.W. (2013) 'Medical conditions in children with Williams syndrome: A systematic review', *Acta Paediatrica*, 102(2), pp. 178–184.

Collins, R.T. (2018) 'Cardiovascular disease in Williams syndrome', *Circulation*, 137(21), pp. 2195–2197.

Davies, M., Udwin, O. and Howlin, P. (1998) 'Adults with Williams syndrome: Preliminary study of social, emotional and behavioural difficulties', *British Journal of Psychiatry*, 172(3), pp. 273–276.

Ewart, A.K., Morris, C.A., Atkinson, D., Jin, W., Sternes, K. and Spallone, P. (1993) 'Hemizygosity at the elastin locus in a developmental disorder, Williams syndrome', *Nature Genetics*, 5(1), pp. 11–16.

Fisher, M.H. and Morin, L. (2017) 'Addressing social skills and problem behaviours in children with Williams syndrome', *Journal of Autism and Developmental Disorders*, 47(3), pp. 835–846.

Hocking, D.R., Bradshaw, J.L., Rinehart, N.J. and Enticott, P.G. (2015) 'Motor development in Williams syndrome', *Developmental Medicine and Child Neurology*, 57(3), pp. 225–232.

Järvinen, A., Korenberg, J.R. and Bellugi, U. (2013) 'The social phenotype of Williams syndrome', *Current Opinion in Neurobiology*, 23(3), pp. 414–422.

Jones, W. et al. (2000) 'Hypersociability in Williams syndrome', *Journal of Cognitive Neuroscience*, 12(Supplement 1), pp. 30–46.

Lucy Pitt (2026) *Williams–Beuren syndrome: signs and possible symptoms which may be observed* (infographic, created 4 May 2026).

Martens, M.A., Wilson, S.J. and Reutens, D.C. (2008) 'Research review: Williams syndrome: a critical review of the cognitive, behavioural, and neuroanatomical phenotype', *Journal of Child Psychology and Psychiatry*, 49(6), pp. 576–608.

Mervis, C.B. and John, A.E. (2010) 'Cognitive and behavioural characteristics of children with Williams syndrome', *Developmental Disabilities Research Reviews*, 16(2), pp. 98–109.

Mervis, C.B. and Velleman, S.L. (2011) 'Children with Williams syndrome: Language, cognitive and behavioural characteristics and their implications for intervention', *Perspectives on Language Learning and Education*, 18(3), pp. 98–107.

Morris, C.A. (2017) 'Williams syndrome', in Adam, M.P. et al. (eds.) *GeneReviews*. Seattle: University of Washington. [Online] Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1249/> (Accessed: 4 May 2026).

Pober, B.R. (2010) 'Williams–Beuren syndrome', *New England Journal of Medicine*, 362(3), pp. 239–252.