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SCHOOL OF HUMANITIES
& SOCIAL SCIENCES

GOLDENHAR SYNDROME

A guide for new parents and families

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ALIGN Quality of Life In Goldenhar Syndrome study



GOLDENHAR SYNDROME

A guide for new parents and families

This guide has been written for new families from a parental perspective. It is based on the views and outlooks of Goldenhar parents, adults with Goldenhar and a small cohort of teenagers. The guide aims to provide information, practical advice, hope and reassurance for families. It may also be useful for medical professionals, educational practitioners and the general-public.

As many of us know, being a new parent is exciting and challenging, but when a child is born with medical complications, additional needs and physical differences (which can be visible and invisible) this can present additional challenges and stress for new families. Please be assured that you are not alone, you are not the only one this has happened to, you are not to blame for these unexpected complications, and the future can be much brighter!

Just like any other child, a child with Goldenhar has every possibility of being able to live a fulfilling and enjoyable life and they have every chance of growing into a healthy, happy and successful adult, able to have a career and life they choose for themselves. There are many success stories of Goldenhar adults who are graduates, authors, graphic designers, digital creators, lecturers, musicians, business developers and owners; and many people who have gone on to have families of their own. There are no limits to what your Goldenhar child could achieve in life!

What exactly is Goldenhar Syndrome?

Goldenhar Syndrome is an 'umbrella' descriptor for a wide range of soft tissue and bone abnormalities which affect the face, the vertebrae and body parts. 70-80% of Goldenhar cases affect one side of the body and face.

Goldenhar shares links with: Hemi-facial Microsomia (HFM); Facio-Auricular-Vertebral-Spectrum (FAV) and Oculo-Auricular-Vertebral-Dysplasia (OAV).

Goldenhar is rare and a unique combination of different conditions put together. Goldenhar prevalence is reported to be 1 in 25,000- 45,000 births. However Hemi-facial Microsomia alone is far more common, occurring in approximately 1 per 6,000 births, HFM can be a stand-alone condition. Many people with Goldenhar have some degree of HFM in combination with OAVs or other unique body differences.

The initial shock – what causes Goldenhar? How do I make sense of Goldenhar Syndrome?

The shock of having a child born with Goldenhar Syndrome can be very traumatic for new parents. Unexplained causation can also be extremely difficult to deal with. It is very important for parents to understand that there is no known cause for this rare syndrome and there are currently no known ways of preventing Goldenhar occurring.

Like many other conditions, Goldenhar Syndrome, occurs sporadically without explanation. Over the years, over 200 environmental and life-style related suggestions have been investigated, in very small studies, none of which have been proven. Genetic research studies are continuing to identify numerous associated chromosomal changes and gene mutations. Goldenhar is complex and a collection of conditions, making it unique to the individual, therefore it is genetically more difficult to pinpoint as the combination of genetic mutations is likely to differ between individuals. Far more research is required.

We appreciate how confusing this is, and how unhelpful the lack of clarity is. Please be assured that parents are NOT to blame for this unforeseeable condition. Goldenhar children are born at random which is why new parents are often shocked and suffer misplaced guilt. Many families find strength from connecting with others 'in the same boat'.

Up until recently Goldenhar was not detected in pregnancy, but recent advancements in scan-technology is helping to identify Goldenhar and other conditions earlier. For future family planning, it is important to understand that it is very unlikely that a family would have another child with Goldenhar.

So, what about my child?

Children born with Goldenhar are individually unique and the spectrum is broad. Goldenhar causes physical differences in the face and body, it is important to note that Goldenhar does not affect intellect or academic ability. Hearing loss is a common feature of Goldenhar and support is often required to avoid communication and learning barriers.

Children may experience any number of the differences listed below. Currently, there aren't any Goldenhar Syndrome medical specialists so you will need to see specialists in each area.

Key areas for you and doctors to look out for:

- Ear deformities, skin tags or absence of an ear.
- Facial asymmetry: one smaller side (hemi-facial microsomia).
- Hearing loss and absence of a middle ear canal
- Mouth/ jaw abnormalities e.g. wide mouth, cleft lip and palate
- Eye: malformations, epibulbar dermoids, absence of an eye.
- Spine: vertebral abnormalities e.g. missing vertebra, missing ribs, a short neck.
- Heart: cardiac defects
- Breathing: respiratory problems
- Urogenital: Ectopic or absent kidneys, missing or additional reproductive organs.

What medical investigations will my child have?

It is likely that your child will have, a hearing test, an eye assessment, an assessment of the face and palate (to check feeding, swallowing and speech), an X-ray of the whole body and spine and an echocardiogram (ultrasound scan of the heart).

Sometimes medical professionals will recommend surgery or medical interventions for improved function or aesthetic reasons. Medical knowledge of Goldenhar is growing slowly and a wide range of specialists will help with the complex aspects of this syndrome. You will receive on-going, professional medical support as your child progresses throughout childhood.

How may these medical interventions affect our lives?

Appointments, medical interventions and surgery are likely to be part of your child's growing up experience. Your child may miss some school due to these on-going appointments and procedures. Juggling appointments, coping with time lost at school and work, preparing for surgery and the recovery, can be challenging for families. If surgical differences are visible, if there are scars or there is swelling from surgery sometimes people may stare out of curiosity. Staring can sometimes make us feel uncomfortable, but remember people are only doing a double-take because they see something different and they are curious. Sometimes a short explanation about the difference, and then a re-focus on similarities can be very helpful. Connecting with other families who have been through similar journeys can help guide you through ups and downs and give you the confidence to move forwards positively.

Educational advancements

R.J Palacio's famous book *Wonder* features a boy with characteristics of Goldenhar Syndrome (OAVs & HFM see p104). *Wonder* aimed to highlight the challenge of living with a visible difference and aimed to provide representation for children living with differences. Palacio's book was made into the famous blockbuster film, *Wonder*, starring a boy with Treacher Collins Syndrome, which shares some parallels with Goldenhar.

Wonder has been hugely influential in raising awareness of what it is like to live with visible differences. A successful campaign by the charity Changing Faces, led to the inclusion of face equality in the National Curriculum and the creation of their 'Wonder' teaching resources which are currently being used in schools. *Wonder*'s educational materials can be found at: <https://www.changingfaces.org.uk/education/education-resources/teachers/classroomresources>



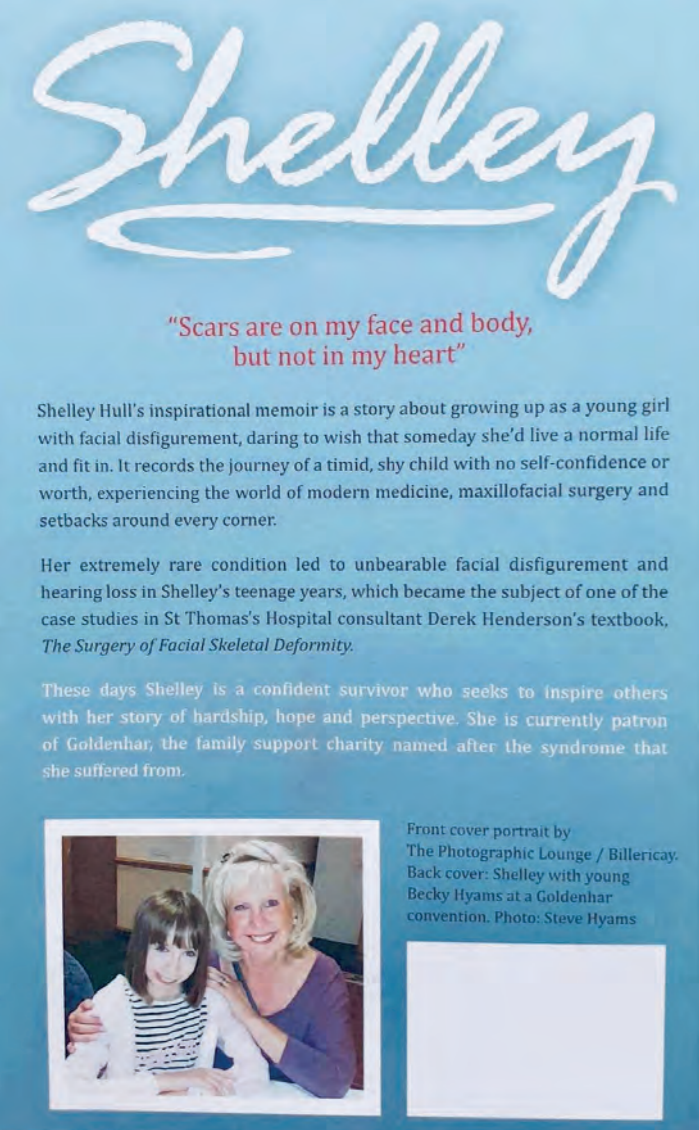
Meet Lucia – child life stories

Lucia has had two operations on her face to repair a cleft-lip, remove skin tags and to repair bilateral macrostomia (a wide mouth). Lucia wears a bone anchored hearing device for hearing loss and has had an operation for this. She also has some differences in her body which can't be seen. She attends regular hospital appointments for her hearing, her spine, her teeth, and feet. Lucia is a happy, outgoing girl, and she lets nothing hold her back! She enjoys art, singing, dancing, baking, karate and socializing with friends. Lucia won her primary school, whole-school talent contest twice; in year 5 for her fantastic singing performance of Lucas Graham's song, Seven Years, and in Year 1 for her dance routine to Roar by Katy Perry. Lucia is excelling in all school subjects and has been studying *Wonder* at school.

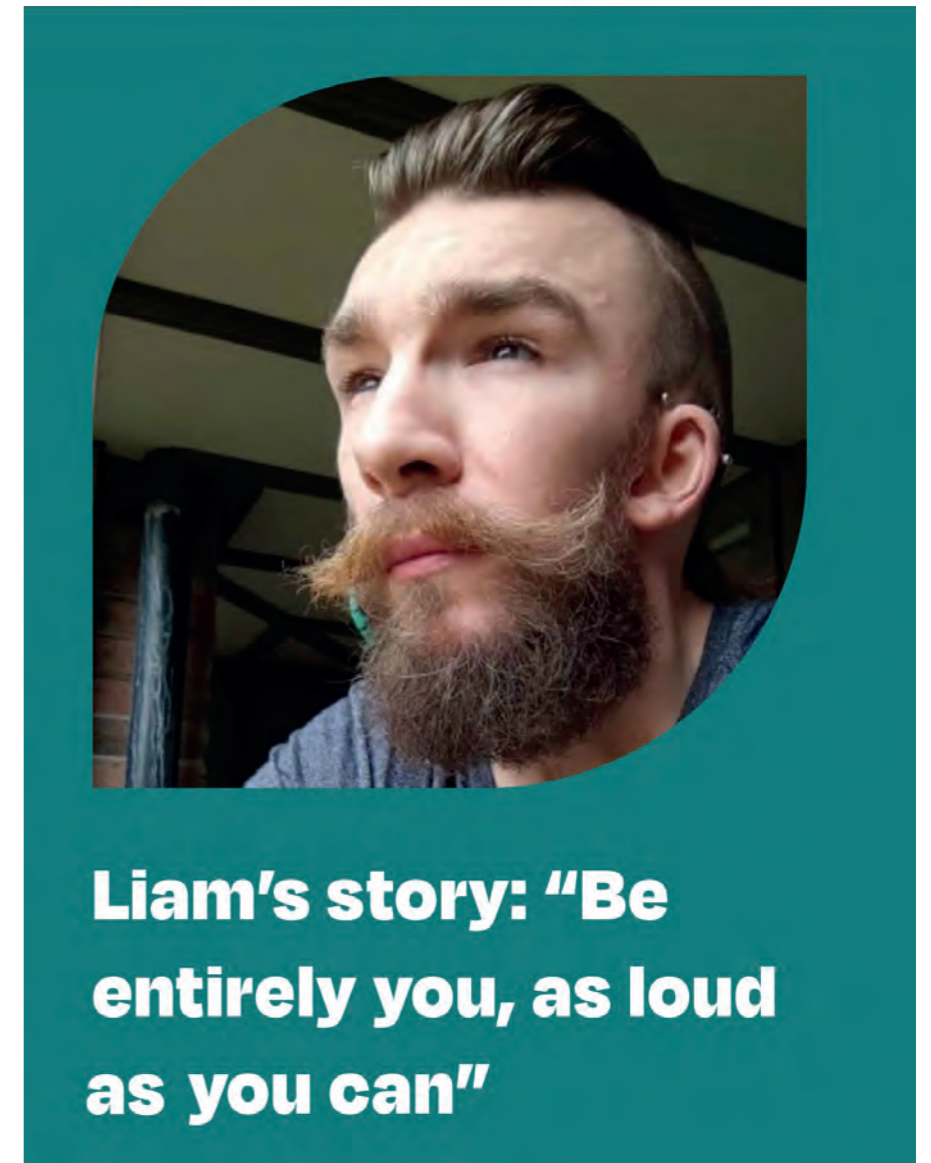


Goldenhar adult life stories

Shelley Hull's inspirational book tells a personal story of what life was like growing up with Goldenhar and aims to provide hope to others living with the Syndrome. Shelley is the patron of the charity, Goldenhar UK, and aims to inspire and support anyone who has ever questioned their self-worth.



Providing support and promoting respect for everyone with a visible difference



The charity, Changing Faces, provides support to individuals with visible differences. Other inspiring Goldenhar life stories can be found on Changing Faces's site.

<https://www.changingfaces.org.uk/story/liams-story-be-entirely-you/>

Goldenhar research: parental perspectives, how might parents feel?

An interview study of ten Goldenhar parents from Goldenhar UK, supported by Leeds Beckett University in 2017, discovered that many new parents reported initial feelings of 'shock', self-blame', 'grief' and 'disbelief. Many parents also said they felt overwhelmed with all the new medical information. Please be assured that these initial reactions are common and normal.

Many parents described how their feelings have changed with time and support. In the study, many parents explained that Goldenhar, had 'enriched' their lives and changed them for the better. Here are some quotes from the study illustrating how parents felt:

The study discovered Goldenhar UK was a valuable support network.

'They have been a massive help with information. It gives me a very good look from where my boy is at - to where he might be going in the future, for his health and stuff!

"It has helped me a lot ...it has helped me feel differently about having a child with Goldenhar."

"We have all been going through a similar sort of journey... we all know the ups and downs.... peaks and troughs of operations.appointments...it is nice to know you've got other families in the same boat and you can talk...it means a lot."

Parents also felt Goldenhar UK was a valuable source of support for children and young people with Goldenhar

"Seeing her build up friendships as well as the friendships we've built was really beneficial it has been really lovely to know that she gets a lot of support out there which in-turn gives us support."

"These events are particularly good for siblings.. my daughter really enjoys coming to these events... she's excited, she's made friends with other siblings, and also a little girl with Goldenhar."

Follow up studies by Leeds Beckett and supported by the Sir Halley Stuart Trust, interviewing Goldenhar adults and teenagers about their quality of life and life outlooks are currently underway.

Here are a few extracts from interviews with adults and young people:

"I've got friends and ..they respect me... as I've got older..I've felt it's become less and less of a big thing for me".

"it doesn't really impact me, just impacts me when it's a loud environment or I've got someone sitting to my left. I usually just tell them to prod me until they get my attention. I learnt over time never to be afraid of telling someone I can't hear."

Adults and young people found the Goldenhar UK weekends beneficial.

"Finding Goldenhar UK.. actually getting to meet people like me that..without any thought or feeling was enough validation to... let me go back and kind of deal with everything."

" It's such a relief...especially when you are younger..you see other kids have like the same thing as you and ..it just shows it's not a big deal. Seeing someone older than me with it the first time was amazing"

"Goldenhar UK for a child is the best start because it gives them support. It gives them the chance to meet other children. It gets them to have fun and actually be themselves. From the parent's point of view it's great for parents to meet other parents. I got good vibes from everybody!"

Here are a few teenage extracts:

"I'm good socialising with people... I try and make myself go out there and talk to people, make conversation."

"I am lucky I've got a really supportive family, really supportive friends. I think you can just sit there and ...think why me? Why me? But that's not gonna fix anything.., that's the way I see it."

"I haven't seen any actors and with Goldenhar on the TV or anything but I really feel like there should be..so you .. create that kind of diversity atmosphere that we should all be different."

Goldenhar UK weekends were described by teenagers as helpful.

"In Chester..... It was pretty good to hear Evie's and Rosa's stories .. they've had loads of surgeries and they've been through so much and it just reflects on... how much I've been through and that there is more people that have been through the same."

"it has definitely made a difference...growing up around other people that experience the same things."

"I like what's going on.. like the other people, and like staying with the kids.. they've got the same...disability ..they can understand .. the difficulties of living with that disability. I do like the whole concept of it."

Here is the link to the parent's study; Breaking the silence:
A qualitative exploration of parental perspectives of children with Goldenhar Syndrome :
<https://doi.org/10.1016/j.heliyon.2024.e24328>



Chris Steele,
former patron of
Goldenhar UK



Goldenhar UK events

Goldenhar UK have first-hand experience of this condition and they can provide practical help, support and advice to families. They offer enjoyable experiences and the opportunity to meet other families at Goldenhar UK family weekends and conferences. Over the years, families have had a lot of fun at Goldenhar UK weekends and many life-long friendships have been formed.

Fundraising has made Goldenhar UK family events possible. Over the years many fundraisers have kindly given up their time and energy to organise fund raising events such as runs, hikes, walks, bike rides, music events, black tie balls, duck races and cake stalls and more to support the charity, Goldenhar UK, and keep it going.

Thank you for reading this guide, I hope you found it helpful and wish you all the best on your new journey.







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Charities and organisations that offer support:

Goldenhar UK: A national support group offering support, advice and guidance for you and your family. Goldenhar UK can offer peer support, conference workshops and family social events.
Contact: www.goldenhar.org.uk

Find them on:  

Changing Faces: this charity supports people who have conditions or injuries that affect their appearance. They offer workshops, resources, a wellbeing support service, support in schools and at work, online advice and a self-help guide.

Call **0345 450 0275**,
Visit www.changingfaces.org.uk/adviceandsupport

Find them on:  

NHS Talking Therapies: Details of local NHS Talking Therapies, for anxiety and depression services are available on the nhs.uk website:
Find an NHS psychological therapies service.

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Sir Halley
Stewart Trust