Unit 1 Maida Vale
Business Centre
Mead Road
Cheltenham
Glos. GL53 7ER

T: +44 (0) 242 261332
M: +44 (0) 7774 465712
www.edsociety.co.uk
E: diana@edsociety.co.uk

International Conference for Ectodermal Dysplasia

9th INTERNATIONAL CONFERENCE FOR ECTODERMAL DYSPLASIA

10th – 12th JUNE 2025

Austin Court, Birmingham, UK

Conference Programme – Session Summaries

Introduction / Context

The 9th International Conference for Ectodermal Dysplasia (ICED25) was held in June 2025 at Austin Court, Birmingham, UK. The event brought together over 80 participants each day, including families affected by Ectodermal Dysplasia, clinicians, researchers, and international support groups from more than 20 countries.

Because ED is such a rare condition, many families and professionals rarely meet others with direct experience. ICED25 provided a unique opportunity for knowledge-sharing, collaboration, and building connections across disciplines and borders.

Aims and Themes

The conference was designed to:

- Strengthen global collaboration on ED care and research.
- Provide families with direct access to medical expertise and practical support.
- Build international partnerships and improve care pathways.

The programme covered a wide range of topics, including genetics, dermatology, dentistry, paediatrics, psychosocial support, and patient advocacy.

Presentation Highlights

Genetics, Taxonomy and "Other Types" of ED

This session provided an update on the classification of ectodermal dysplasias and advances in understanding the EDA pathway. Presentations highlighted structural modelling of gene–protein interactions, new insights into rare subtypes such as AEC syndrome, and updates on less common genetic contributors. The discussions emphasised the importance of accurate taxonomy in guiding diagnosis, treatment, and research collaboration.

Dental Implants and Oral Health

Dental care was a major theme, with focus on innovations in dental management, results from the international ED oral health study, and findings from a UK survey on dental implants by age and region. Presentations explored how early intervention, access to implant services, and shared data can improve outcomes. The session closed with a broad discussion on best practice in treatment planning and patient care.

Thermoregulation Throughout the Lifespan

Talks explored the challenges of insufficient thermoregulation in infants, differences in heat regulation between people with ED and controls, and the impact of thermoregulation difficulties on sleep quality. Practical strategies for cooling and managing temperature across age groups were also shared. The session underlined how critical thermal management is for health, quality of life, and daily functioning. The implications of global warming and climate change for those with the hypohidrotic ectodermal dysplasias were mentioned.

Dermatology

This session examined how ED affects hair, nails, and skin across different subtypes. Updates ranged from the biology of hair and nail growth to the wider dermatological manifestations that can impact daily living. The talks highlighted the importance of dermatology within multidisciplinary care, both for diagnosis and for improving quality of life.

Psychology and Social Science

The psychological and social impact of ED was explored from multiple perspectives. Presentations addressed lived experience, face equality, family support needs, and the impact of stigma on reproductive decision-making. Broader themes of psychosocial wellbeing in rare disease communities were also covered, reinforcing the need for holistic, person-centred care.

Submitted Abstracts

Researchers and clinicians shared a series of short presentations covering topics such as atopy in ED, the experiences of female carriers of XLHED, saliva studies, rare case reports, the role of paediatric dentistry, and examples of multidisciplinary dental rehabilitation. Additional insights included healthcare experiences and barriers faced by people living with ED, providing valuable real-world data to inform future services.

Clinical Services / Multidisciplinary Teams

This session focused on models of multidisciplinary care, ENT involvement in ED, and the critical issue of transition from paediatric to adult services. Updates also included emerging clinical guidelines for specific genetic subtypes. The discussion stressed the importance of coordinated lifelong care pathways tailored to the complex needs of ED patients.

Trials, Treatments and Databases

The final scientific session reviewed progress in clinical trials, including the EDELIFE trial, advances in predicting phenotype from genotype, and the role of international databases in research. Presentations also covered prevalence rates of ED and the broader public health perspective of rare diseases, with an emphasis on improving support and care for affected families worldwide.

Family Day

The third day of ICED25 was dedicated to families. Presentations were delivered in accessible language, focusing on practical advice and everyday challenges. Families valued the opportunity to ask questions directly to a panel of experts covering dentistry, dermatology, genetics, and paediatrics. There was also a session specifically on problems caused by changes in the *TP63* gene.

For many, it was the first time meeting another person with ED. The day created a strong sense of community, reducing feelings of isolation and empowering families to share knowledge and support one another.

Overall Reflections

Across all sessions, ICED25 showcased the remarkable progress being made in both scientific research and clinical care for ectodermal dysplasias. The programme highlighted advances in genetics, innovative treatment trials, and the growing role of international databases in driving collaboration. At the same time, there was a strong emphasis on lived experience, psychology, and the importance of patient-centred support.

Themes of multidisciplinary teamwork, early intervention, and the need for coordinated lifelong care ran throughout the conference. The breadth of topics—from taxonomy to dental health, dermatology to psychosocial wellbeing—demonstrated that addressing ED requires not only medical expertise but also social awareness, family support, and a global commitment to collaboration. ICED25 not only provided new knowledge but also strengthened the international community of researchers, clinicians, and families working together to improve the lives of those affected by ED.

The impact of the conference was clear. Families reported feeling more informed, supported, and connected, while clinicians gained valuable insight into the lived experiences of ED, an aspect often absent from clinical training. The event also led to the establishment of new international collaborations, paving the way for joint research, clinical trials, and shared resources. Importantly, the Society further strengthened its reputation as a global leader in ED support and advocacy, ensuring that the voices of those affected are represented at the highest level.

Individual Summaries

Session One – Genetics and Taxonomy

What Is and Isn't an Ectodermal Dysplasia?

Nicolai Peschel (Erlangen, Germany)

Ectodermal dysplasias (EDs) are a diverse group of congenital genetic disorders affecting structures derived from the ectoderm, including hair, teeth, nails, and certain glands. Initially identified through clinical observations in the 19th century, these conditions were later classified systematically by Newton Freire-Maia in the 1970s, based on anatomical features and inheritance patterns. With advances in genetics, modern classifications now focus on molecular pathways and genes.

The current classification system divides EDs into six major groups based on the signaling pathways affected. Group 1 involves mutations in the ectodysplasin A (EDA)/NF-κB pathway, leading to conditions such as hypohidrotic ectodermal dysplasia. Group 2 is characterized by abnormalities in the Wnt/β-catenin signaling pathway, affecting tooth and hair development. Group 3 includes EDs caused by disruptions in the p63 transcription factor network, often resulting in limb and facial abnormalities. Group 4 comprises disorders affecting structural proteins involved in keratinization and cell adhesion. Group 5 includes conditions with defects in other signaling pathways, such as Notch signaling, while Group 6 consists of syndromes with partial ED-like features but affecting non-ectodermal tissues.

As genetic research continues, classifications are expected to evolve, incorporating newly discovered pathogenic variants and refining disease groupings. Understanding the molecular basis of EDs not only aids diagnosis but also facilitates targeted therapeutic approaches.

Session One - Genetics, Taxonomy and "Other Types" of ED

Molecular Aspects of the EDA Pathway and Ectodermal Dysplasia

Denis Headon (Roslin, Scotland)

Hypohidrotic Ectodermal Dysplasia (HED) is a developmental condition characterised by absent or reduced sweat and meibomian glands, sparse hair, missing or differently shaped teeth, and reduced salivary, respiratory, and mammary glands. Mouse models of HED, first identified and described in the mid-20th Century, replicate the inheritance and clinical features of the human condition. These mouse models were instrumental in identifying the causative genes EDAR and EDARADD, and in uncovering the developmental processes disrupted in HED.

Studies in both mice and humans reveal that many of the structures affected by HED begin their development as solid cords of cells that grow down from the embryonic epidermis (ectoderm). The EDA pathway is central to this process: EDA is a secreted extracellular molecule that contacts the receptor EDAR on the surface of epithelial cells, which then signals through the adaptor protein EDARADD and a series of other proteins to activate the transcription factor NF-κB. This relay of signals from one protein to the next promotes the full development of ectodermal structures. Because NF-κB is used by cells in many organs, HED can, in some rarer cases, also form part of broader syndromes affecting the immune system and the skeleton.

Understanding this pathway has several uses. It provides the genetic basis for diagnosis and counselling, clarifies the cell and developmental origins of HED phenotypes, and opens a therapeutic avenue. Since EDA is a protein that works outside of cells to stimulate the EDAR on their surfaces, it is relatively easy to deliver to the body. However, this delivery will only be effective if given in the appropriate time during early development. This approach has shown promise in mouse models and is now being trialled in humans.

Despite progress, many questions remain. Why is the embryonic response to EDA limited to a narrow time window in development, and can this window be extended or reopened? Why do some structures require EDA to form while other closely related structures do not? Exploring these issues, particularly the role of the deeper mesenchymal cells, may help guide therapies beyond EDA protein replacement. Such insights are also relevant to other conditions, including focal dermal hypoplasia, and to regenerative medicine.

Session One - The commonality of TSPEAR-related ARED14 sheds light on human origins

Dr. Adam Jackson (Manchester, England) discussed autosomal recessive ectodermal dysplasia type 14 caused by biallelic variants in *TSPEAR* (a gene that encodes a protein of unknown function).

Overview of Presentation:

- Hypodontia and conical-shaped teeth are the predominant features of ARED14
- Missense variants (that alter the chain of amino acids in the protein) perturb the beta propeller formed by several EAR domains in TSPEAR
- Multiple founder variants in TSPEAR, originating at the end of the Last Glacial Maximum, are responsible for ARED14 in non-Finnish Europeans with up to 1 in 140 of these individuals being carriers for ARED14.
- TSPEAR shows restricted expression to ectodermal placodes in keeping with its Drosophila ortholog, Closca, which is well known to coordinate extracellular matrix (ECM)-dependent signalling.
- Knockout of TSPEAR orthologs in zebrafish recapitulates the human dental phenotype whilst also demonstrating a possible role for TSPEAR in WNT signalling in fin regeneration
- TSPEAR's macromolecular structure makes it a likely candidate for WNT ligand sequestration in the ECM.

Session Four - Hair in the Ectodermal Dysplasias

Andrew Messenger (Sheffield, England)

Embryonic development of hair follicles involves a cascade of chemical signals passing between cells of the fetal epidermis – the outer ectodermal layer of the skin – and the underlying cells that will form the dermis. Ectodermal dysplasias involving hair growth are due to alterations (variants or mutations) in genes that control these signals, resulting in poor or absent hair follicle development. The development of the growth phase of the adult hair cycle, known as anagen, involves similar chemical signals, hence gene variants that impair hair follicle development can have ongoing effects on hair growth throughout life.

Most types of ectodermal dysplasia show reduced hair growth on the scalp (hypotrichosis). This may vary from mild thinning to almost complete absence of hair growth. Hair growth in other sites may also be sparse including eyebrows, eyelashes and body hair. The texture of the hair is variable. In HED forms hair fibres are variously described as fine in calibre, soft and silky, wiry and light in colour. The hair in AEC and EEC syndromes may be more brittle and wiry and with some patchy loss of hair, and fine and brittle in Clouston syndrome. Hair loss in incontinentia pigmenti (IP) follows the linear streaky pattern where skin was inflamed in the neonatal period and may represent loss of hair follicles in these sites. It typically affects the crown region in adults. The hair in IP may also be coarse and wiry.

The clinical diagnosis of ectodermal dysplasia in those presenting with hypotrichosis rests on signs of abnormalities in other ectoderm-derived tissues (teeth, nails). With the possible exception of IP, the hair abnormalities in ectodermal dysplasias are not specific and there are other genetic hair and acquired hair disorders that present with sparse hair.

There are no treatments that will correct the underlying cause of impaired hair growth in ectodermal dysplasias. Treatment with topical or oral minoxidil may help to improve hair growth to some extent

although the response is variable and treatment must be continued indefinitely to maintain it. Hair transplant surgery can be helpful in cases where hair loss is localized, such as may occur in IP. However, hair transplantation is not suitable for the majority of people with ectodermal dysplasias where hair growth is generally sparse. The appearance of hair may be improved by simple hair care measures that improve texture and reduce hair breakage. Some people find aids such as wigs or weaves helpful. Further support may be needed in those who struggle to cope with the psychosocial aspects of a lack of hair. This can come from a number of sources – family, GP, school nurse, psychological services and peer groups. Alopecia UK (https://www.alopecia.org.uk) is devoted to helping those with hair loss from any cause and can be an invaluable source of information and help on coping with the physical and emotional consequences.

Session Two – Dental Implants and Oral Health
Clark Stanford DDS (Iowa, USA)
New Approaches in Dental Management of the Ectodermal Dysplasias

In the past few years there has been significant changes in dentistry that supports the care of folks with ectodermal dysplasias. The most significant is the imaging and software platforms allowing a patient and their care team to predict the outcomes of care and compare between different options before anything is done. The use of bone and soft tissue grafts is evolving as implant designs are becoming smaller and other areas of the jaws are used to support teeth for those who did not form teeth. There are regional differences in approaches based on health care systems and resources. It was reviewed that there are always multiple options from those that are more conventional approaches (e.g., partial or complete dentures or overdentures on remaining teeth) that are very appropriate, to conservative bonded bridges (so called “Maryland” bridges) to the use of various forms of oral implants. Patients have multiple options and should advocate for a range of choices with their hospital or provider since there is never one solution, there are always options. The wants, needs and desires of the patient and family are key aspects for successful outcomes of care.

Session Three - Thermoregulation throughout the lifespan
Heather Massey (Portsmouth, England)
Thermoregulation in Ectodermal Dysplasia

Some people with ED lack functioning glands, including sweat glands, which affects their capability to regulate their body temperature (thermoregulation), particularly in warm conditions. While sweating is a key method of heat loss, the body can also regulate temperature by increasing blood flow to the skin (dry heat loss). However, previous research suggests this mechanism may also be impaired in some people with ED.

In our study, 16 individuals with ED and 9 matched (for height and mass) controls exercised in a warm environment while we monitored body temperature, skin temperature, sweat rates, and skin blood flow. Cooling interventions were available if needed. The goal was to assess thermoregulation and identify effective cooling methods.

All participants with ED could redistribute blood to the skin to support dry heat loss. Some could also sweat adequately; others showed uncertain results, while two (with X-linked ED) could not regulate their temperature.

We found that cooling through hand, arm, or foot immersion in cold tap water—or using water sprays and fans—was effective. Future research should explore which genetic diagnoses influence thermoregulation (as well as sweating) and if participation in regular physical activity or heat acclimatisation can improve sweat production and temperature control.

Session Five - Psychology and Social Science.
Phyllida Swift (Face Equality International)
An Introduction to the Work of Face Equality International

Phyllida Swift presented on the work of Face Equality International. As an Alliance for face equality, FEI works to ensure the global facial difference community are respected, protected and thriving. Together with their members, FEI runs International Face Equality Week each year, whilst running an educational programme for non-profits to build their capacity for societal change. It's FEI's vision to create a society free from barriers for the FD community. Find out more about their work via their social channels on @faceequalityint or faceequalityinternational.org.

Session Six - Submitted Abstract Presentations
Laura Krogh Herlin (Aarhus, Denmark)
Nationwide study on atopic disorders in patients with ectodermal dysplasias

Ectodermal dysplasias (EDs) - especially hypohidrotic ED - have previously been linked to a higher risk of allergic (atopic) conditions in survey studies and case series. However, larger population-based studies have been missing.

In our study, we used national health records from Denmark to identify and follow a cohort of nearly 400 patients diagnosed with ED and compared them to individuals without ED. We investigated how often they were diagnosed in hospitals with atopic dermatitis (eczema), asthma, hay fever, and food allergies. We also examined prescriptions for allergy medications to get a better understanding of atopic diseases treated outside hospital settings. We obtained linked hospital and prescription data from national health registries.

Our results show that people with hypohidrotic ED, especially males with the X-linked hypohidrotic ED, are significantly more likely - about 6 to 8 times - to be diagnosed with allergic conditions. The relative increase in risk was highest for atopic dermatitis. In contrast, we did not see the same increased risk in women with X-linked hypohidrotic ED or in people with other types of ED. The increased risk for atopic disorders in hypohidrotic ED was further confirmed in secondary analyses using prescription data.

These findings highlight the importance of monitoring for atopic conditions in individuals with hypohidrotic ED and ensuring they receive timely and appropriate care to manage symptoms and improve quality of life.

Isabelle Luchsinger (Zürich, Switzerland; formerly Paris, France)
Clinical Characterisation of Women Carrier of X-linked Hypohidrotic Ectodermal Dysplasia (XLHED)

X-linked hypohidrotic ectodermal dysplasia (XLHED) is the most common form of ectodermal dysplasia. It is caused by changes in the *EDA* gene on the X chromosome, so men usually show a consistent pattern of symptoms, while women are often described as "carriers" with only mild signs. However, our study of 36 adult women (30 of them mothers) shows that many women are significantly affected.

Almost all women had differences in their teeth. Nearly half of the cohort were diagnosed with six or more missing teeth. Many women had problems with sweating. Five women could not sweat at all. Most other women had reduced sweating, sometimes only on one side of the body. Some women who had gone through menopause started sweating for the first time in areas where they had not been able to before.

Breast development was frequently affected. Two women had no breast tissue at all, one had only one breast, and more than half showed significant size differences. Some patients also had small or inverted nipples, extra nipples, and reduced nipple glands. Of the 30 mothers, only four could breastfeed without difficulty. This led to emotional distress after childbirth.

Changes in hair growth were another major concern. Three-quarters of the women had thinning of the eyebrow parts on the sides. A majority showed hair loss on the scalp in characteristic linear pattern. Dry eyes and mouth were reported as often as in male patients.

In the era of prenatal therapies, the timely recognition and diagnosis of affected females is becoming increasingly important. Clinical clues may include linear alopecia, thinning of the lateral portions of the eyebrows, asymmetrical sweating and flat nipples.

Overall, this study shows that women with XLHED can have significant health issues and should not be considered only as symptom-free “carriers.” They would benefit from regular, adapted interdisciplinary medical care similar to male patients.

Amanda Swanson (Columbus, Ohio, USA)
“Rare Diseases and Public Health: Improving Care for Families Affected by ED”

This mixed methods research study explored how individuals with ED and their families experience the healthcare system, including both the challenges and what helps care go more smoothly. The study involved a US-based national survey and focus groups with affected adults and caregivers of children with ED.

The biggest difficulties were finding knowledgeable providers, coordinating care between specialists, and dealing with insurance – especially for dental care. Despite these hurdles, families showed great determination, often traveling long distances and doing much of the coordination themselves. Positive experiences frequently resulted from interactions with providers who were open-minded, willing to learn, and treated families as partners on the care team. Emotional and mental health impacts were also common, including stress and worry - but also strength and resilience, especially when families found supportive communities through social media groups and ED organizations.

Oral health stood out as a top concern, with families emphasizing how missing teeth can affect eating, speaking, confidence, and overall quality of life. Advocacy was a strong theme, referring to both self-advocacy and on a broader policy level, with many families working hard to secure accommodations and push for better access to care.

This research highlights the critical need for more coordinated, compassionate care and dental coverage that recognizes the medical necessity of treatment. It also reinforces the power of patient and caregiver voices in shaping more inclusive healthcare practice, and the importance of community in building resilience for those living with rare diseases like ED.

Session Seven - Clinical services/MDTs
Tess McPherson (Oxford, England)
Transition from paediatric to adult services

Adolescence is a time of rapid change and growth and specific challenges to managing chronic conditions. Young people living with ED face specific challenges including symptoms such as itch and photo sensitivity, appearance related concerns and impact on self-esteem and mental health.

This talk covered ways to communicate with young people with both the experience of a specialist service for young people with skin conditions that integrates psychological support and tips to use in all settings to support young people living with ectodermal dysplasia's, including impact and solutions to support young people through teenage years and into adulthood. 2 cases were presented with specific issues with skin, photosensitivity, dental issues and breast development. Support for mental health guidance was presented on assessment and support for mental health conditions in children and young people with skin conditions.

Family Day Session One (Focus on p63: EEC, AEC, etc.)
Mario Vincenzo Di Iorio Enzo (Padua, Italy)
Advanced ocular therapy in p63-associated disorders

Ectrodactyly-Ectodermal Dysplasia-Clefting (EEC) syndrome is a rare autosomal dominant disease, due to mutations in p63 gene, essential for corneal repair and regeneration (Di Iorio et al.2012). While in childhood, clefting and hand deformities are the main clinical features, during early adulthood the ocular problems become the predominant clinical aspect due to progressive bilateral LSCD, eventually leading to corneal degeneration (Di Iorio et al. 2012). The ocular effects of EEC have a tremendous impact on patients' quality of life and medical and surgical treatments are ineffective or of limited benefit. The most common causes of the condition are the gene alterations R279H and R304Q, and result in the most severe corneal damage. OMESCs represent an alternative source of stem cells capable of regenerating the corneal surface, the epithelium, (Nishida et al. 2004) and could be combined with gene therapy to provide an attractive therapeutic option. We recently identified powerful small interfering RNAs (siRNAs) for the R279- and R304- variants in the p63 gene, with no effects on wild-type p63. Long-term treatment resulted in a population of long-lived cells with extended survival, thus counteracting premature stem cell aging in vitro, and was shown to be effective in correcting the slower epithelial growth, giving rise to a full thickness of stratified and differentiated epithelium. Since the corneal pathology in EEC follows a clear clinical course with LSCD usually manifesting in the 2nd to 3rd decade and leading to severe corneal failure in the 4th-5th decade, this provides a therapeutic window to correct the genetic defect. In early corneal disease, with LESCs still present in the limbus, the use of eye drops containing mutant-specific siRNAs may be an effective therapeutic option. In adult patients with established LSCD and no LESCs, clinical outcomes may be improved by ex vivo gene correction of OMESCs with p63 mutant-specific siRNAs.