ESPE NEWS
BRINGING THE LATEST IN PAEDIATRIC ENDOCRINOLOGY TO YOU

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Special issue
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ESPE 2022 in Rome, Italy – submit your abstracts by 19 April!
The ESPE Rare Disease Advisory Group began its work last year. Its aim is to identify gaps and duplications between work on rare conditions carried out by ESPE and that co-ordinated by others, such as the European Reference Networks, patient organisations and other societies. The Rare Disease Advisory Group has already made good progress, as its Chairs, Faisal Ahmed and Rasa Verkauskiene, report on page 8.

It is the Group’s valuable work that inspired the theme of this issue of ESPE News, which includes the latest developments in a range of rare paediatric endocrine conditions.

On page 5, Asma Deeb looks at the factors underlying disorders of growth plate function. She brings us up to date with the genetic causes of a number of these conditions. As Asma says, the growth plate has a high level of complex machinery critical for normal growth, but is often overlooked in discussions of paediatric endocrine disease.

The genetic basis of Beckwith−Wiedemann syndrome is described by Khalid Hussain on page 6, along with its endocrine effects, the most common of which is hyperinsulinaemic hypoglycaemia. Recent guidelines provide important recommendations for the management of this rare condition.

Rebecca Brown and Melissa Lighthouse consider the treatment of lipodystrophy on page 7. The use of leptin, in the form of metreleptin, reduces hyperphagia, improves insulin resistance and diabetes, and reduces serum and hepatic triglycerides in generalised lipodystrophy. Insulin resistance can improve dramatically, with resolution of diabetes even in patients who required high doses of insulin.

Excitingly, we can now look forward to meeting face-to-face at the 60th Annual ESPE meeting in Rome on 15–17 September 2022. The contributions of ESPE members are a crucial part of the event, so please submit your abstracts by 19 April. Early bird registration ends on 20 June. Find out more at www.eurospe.org/meetings/2022/espe-2022.

I hope you enjoy reading through this issue to find all the other news and information about ESPE activities. Your feedback is always welcome!

Sarah Ehtisham
Editor, ESPE News
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ESPE COVID-19 Hub

Remember: the ESPE COVID-19 Hub is a resource to support physicians and patients and their families during the pandemic.

You can use it to find a wide range of information relevant to paediatric endocrinology and COVID-19.

www.eurospe.org/patients/espe-covid-19-hub

ESPE e-Learning

Supporting this issue’s theme

The following chapters with cases on rare diseases can be found under Courses in paediatric endocrinology and diabetes:

• Adrenal disorders
• Calcium and bone
• Diabetes
• Disorders of sex development
• Growth and growth regulation
• Hypothyroidism
• Intrauterine growth retardation or small for gestational age (average)
• Monogenic diabetes (easy and average)
• Multiple pituitary hormone deficiency (easy and average)
• Puberty and the GH-deficient child (average)
• Type 2 diabetes and obesity (easy)

These are available in English, and some (but not all) have been translated into Arabic, French or Spanish.

News in e-learning

• A new chapter, Caring for gender diverse youth, has been added in Transgender care, under Courses in paediatric endocrinology and diabetes.

Can you help with translation?

If you would like to benefit patients in your region by translating leaflets into your own language (which need not be one of those mentioned above), please contact espe@eurospe.org using the form at the second link below.

ESPE patient leaflets

ESPE provides patient leaflets on the following topics, in easy and/or average readability (as indicated):

• Congenital adrenal hyperplasia (easy)
• Constitutional delay of growth and puberty (average)
• Craniohypophyseal hypoplasia (easy and average)
• Diabetes insipidus (easy and average)
• Growth hormone deficiency (easy and average)
• Hyperthyroidism (easy and average)
• Hypothyroidism (easy and average)
• Intrauterine growth retardation or small for gestational age (average)
• Monogenic diabetes (easy and average)
• Multiple pituitary hormone deficiency (easy and average)
• Puberty and the GH-deficient child (average)
• Type 2 diabetes and obesity (easy)

These are available in English, and some (but not all) have been translated into Arabic, French or Spanish.

You can download the leaflets from www.eurospe.org/patients

To help with translation see www.eurospe.org/news/item/15324

Your chance to develop ESPE e-Learning

There are currently vacancies for the e-Learning Committee Chair and two Committee members. These are perfect opportunities to make a real difference to ESPE’s key aims in education, and would suit anyone with an interest in advancing online education.

Apply by 31 March 2022

Find out more at www.eurospe.org/about/vacancies
Bringing you recent highlights from the world of research

Metabolic co-morbidity in CAH

This longitudinal study by Torky et al. aimed to assess cardiovascular risk factors and metabolic morbidity in patients with classic congenital adrenal hyperplasia (CAH) during childhood and adulthood.

Patients with classic CAH were found to have metabolic morbidity starting at an early age, prior to puberty. Compared with the general US population, children with CAH had a higher prevalence of obesity, hypertension, insulin resistance, fasting hyperglycaemia and dyslipidaemia. Fludrocortisone dose was associated with hypertension in childhood, which was most commonly observed in children <2 years. Adults with CAH had a higher prevalence of obesity, hypertension and insulin resistance. Maternal obesity during childhood was the only contributing factor to adult obesity. Suppressed androstenedione, reflecting excess glucocorticoid therapy, was a contributing factor for hypertension across all ages and suppressed testosterone was associated with insulin resistance in adults.

These findings implicate treatment-related metabolic risk, and call for careful monitoring and judicious use of glucocorticoid and mineralocorticoid replacement in CAH.

Endogenous cannabinoids and MC4R in control of energy homeostasis

Despite major advances in recent years, regulation of food intake and energy homeostasis is not completely understood. As a consequence, there are very few pharmacological therapies for disorders caused by a disturbed regulation of food intake, ranging from obesity to anorexia.

Yong et al. have examined a link between endocannabinoid and melanocortin signalling networks. They used several in vivo and ex vivo approaches to identify the endocannabinoid compound 2-arachidonoylglycerol (2-AG) as a suppressor of GABAergic input into melanocortin-4-receptor (MC4R)-expressing neurones in the paraventricular nucleus of the hypothalamus.

Chemically or genetically suppressing 2-AG synthesis in MC4R neurones led to a reduction in activity of these neurones and, consequently, to reduced food intake and body weight in mice that were also resistant to diet-induced obesity. The 2-AG synthesising enzyme diacylglycerol lipase-α could therefore be a potential target to treat obesity.

BDV: an emerging syndrome of early childhood obesity

Bosch et al. have described a new syndrome, termed Blakemore-Durmaz-Vasileiou (BDV) syndrome, in four affected individuals from three unrelated consanguineous families: two siblings of Syrian descent, one of Egyptian descent and one of Pakistani descent.

The underlying genetic cause was found to be biallelic loss-of-function variants of the CPE gene, which is localised in chromosomal region 4q32.3 and encodes carboxypeptidase E, an enzyme that converts proenkephalin peptides and propeptide hormones to bioactive forms. It is widely expressed in the central nervous system and endocrine tissues, including the adrenal medulla and adipose tissue.

Predominant clinical features of BDV syndrome include severe early childhood obesity, hyperphagia, infantile hypotonia and neurodevelopmental delay. Endocrine abnormalities include hypogonadotrophic hypogonadism, hypothyroidism, insulin resistance and diabetes. Since the overall clinical presentation overlaps with that of Prader–Willi syndrome, BDV syndrome should be considered in the differential diagnosis.

Closed-loop control in very young children with type 1 diabetes

Ware et al. conducted a randomised, cross-over trial over seven centres in four European countries (Austria, Germany, Luxembourg and the UK), including 74 children (5.6±1.6 years) with type 1 diabetes on sensor-augmented insulin-pump therapy. The participants received the Cambridge algorithm closed-loop system (intervention) and sensor-augmented pump therapy (control) in two 16-week periods, randomly. The primary endpoint was the between-treatment difference in the percentage of time in range (70–180 mg/dl), measured by the glucose sensor.

Glucose level in the target range was 8.7% points (95% CI, 7.4−9.9) higher in the closed-loop period than during the control period (P<0.001). The improvement in the percentage of time spent in a hyperglycaemic state, the glycated haemoglobin level, and the mean sensor glucose level were significantly better for the close-loop period (P<0.001 for all). The time spent in hypoglycaemia was similar between the two treatments (P=0.74).
The growth plate: an ignored organ

Asma Deeb examines our increased knowledge of the genetics behind growth plate disorders.

The fast development of technology related to genetic studies, and animal and in vitro experiments on the epiphyseal growth plate (GP), have led to a revolution in knowledge of and genetic discoveries in human growth disorders. Various novel genetic causes of growth failure have now been described, with direct implications for diagnosis and treatment. Many genes have appeared in gene expression studies of the different zones of the GP.1,2

The GP is the structure where linear growth takes place. In the GP, chondrocytes proliferate, hypertrophy and secrete cartilage extracellular matrix, under the influence of endocrine and paracrine factors. The main elements affecting the functional integrity of GP relate to paracrine factors, extracellular matrix, intracellular pathways and fundamental cellular processes.3 A defect in any of these impairs normal GP function and adversely impacts linear growth.

Here, I will discuss some examples of growth disorders associated with these various factors in GP function.

Fundamental cellular processes

Genetic mutations related to fundamental cellular processes can cause severe growth deficiency, affecting both pre- and postnatal growth.4 They can be associated with normal head circumference, such as 3-M syndrome, which is caused by CUL7, OBSL1 or CCD8C.5 Defects in these genes lead to aberrant cell division and growth failure.4

Microcephalic primordial dwarfism is another phenotype in this category. It is also characterised by severe pre- and postnatal growth retardation, but accompanied by microcephaly.6 A further example, Seckel syndrome, is caused by mutations in different genes encoding proteins related to the response to DNA damage.6

Paracrine functions

Paracrine regulation plays a major role in the GP, and most defects result in skeletal dysplasia with disproportionate short stature.7 A well known example in this category relates to fibroblast growth factor receptor-3 (FGFR3), which acts as a negative regulator of GP chondrogenesis. Heterozygous activating mutations in FGFR3 impair bone elongation and result in the hypochondroplasia–achondroplasia spectrum.8

C-natriuretic peptide (CNP) and its receptor provide another example of the effect of paracrine regulation. Homozygous inactivating mutations of NPR2 (encoding the main CNP receptor) cause acromesomelic dysplasia, Maroteaux type 2, which is a severe form of skeletal dysplasia.9 The phenotype of heterozygous NPR2 mutations is similar to that of SHOX haploinsufficiency (Léri–Weill syndrome), with short forearms and lower legs (mesomelia). It is reported that the heterozygous NPR2 mutations may explain 2–3% of cases of idiopathic short stature.10

Extracellular matrix

Chondrocytes at the GP have a unique function of secreting extracellular matrix containing specific collagens and non-collagenous proteins and proteoglycans that are required for normal GP function. Mutations in several genes encoding these proteins lead to several phenotypes of growth disorder, which can be categorised as defects of extracellular matrix.

One of these is brachyolmia, due to mutations in PAPSS2. This gene is required for sulphation of a variety of molecules, including cartilage glycosaminoglycans. Its mutation results in adverse effects on spine and long bone growth.11 Other phenotypes in this category include congenital spondyloepiphyseal dysplasia due to COL2A1 mutations, and geleophysic dysplasia due to ADAMTS12 mutations.1

Intracellular pathways

Various intracellular pathways play a role in chondrocyte differentiation in the GP, and multiple genes encode proteins that are critical here. One example is the EvC gene, a mutation of which causes Ellis–Van Creveld (EvC) syndrome. Animal models have shown that EvC syndrome is a disorder of chondrocyte differentiation, with accelerated differentiation and premature hypertrophy of chondrocytes.12 Smith–McCort dysplasia is another example, resulting from RAB33B mutation, which interferes with Golgi vesicle release at the GP.

In summary, the GP is a fascinating yet ignored organ, that has a high level of complex machinery critical for the normal growth process.

Asma Deeb
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References
10. Wang et al. 2015 Human Mutation 36 474–481.
12. Muscato et al. 2015 Endocrine Pathology 52 957–966.
Beckwith–Wiedemann syndrome: endocrine manifestations

A comprehensive review of the latest understanding of this rare disease is provided by Khalid Hussain.

Beckwith–Wiedemann syndrome (BWS) was first described by the American paediatric pathologist John Bruce Beckwith and the German paediatrician Hans-Rudolf Wiedemann.1

**Genetic basis of BWS**

BWS is an imprinting disorder and the imprinted region is located on chromosome 11p15.5. This region consists of two imprinting domains IGF2/H19 and CDKN1C/KCNQ1OT1. There are five known epigenetic and genetic causes of BWS, and these may be mosaic in different tissues, thus accounting for the variability in the phenotype.2 These include loss of methylation at KvDMR1, gain of methylation at H19DMR, paternal uniparental disomy (UPD), CDKN1C mutations and chromosomal rearrangements. In addition, some BWS patients show multi-locus imprinting defects, with methylation changes extending to other imprinted genes.

**A need for guidelines**

Given the absence of consensual recommendations or international guidelines, the Scientific Committee of the Italian BWS Association and the First International BWS Consensus Group (41 experts from 11 European countries and the USA, including patient support group members) have made recommendations for the diagnosis, molecular testing, clinical management, follow-up and tumour surveillance of patients with BWS.3,4 They have made 72 recommendations for the molecular diagnosis and clinical management of patients with BWS.5

**Table. Cardinal and suggestive features of Beckwith–Wiedemann spectrum.**

<table>
<thead>
<tr>
<th>Cardinal features (2 points per feature)</th>
<th>Suggestive features (1 point per feature)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Macrognathia</td>
<td>Birthweight &gt;2SDS above the mean</td>
</tr>
<tr>
<td>Exomphalos</td>
<td>Facial naevus simplex</td>
</tr>
<tr>
<td>Lateralised overgrowth</td>
<td>Polyhydramnios and/or placental megaly</td>
</tr>
<tr>
<td>Multifocal and/or bilateral Wilms tumour or nephroblastomatosis</td>
<td>Ear creases and/or pits</td>
</tr>
<tr>
<td>Hyperinsulinaemia (lasting &gt;1 week and requiring escalated treatment)</td>
<td>Transient hypoglycaemia (lasting &lt;1 week)</td>
</tr>
<tr>
<td>Pathology findings: adrenal cortex cytomegaly, placental mesenchymal dysplasia or pancreatic adenomatosis</td>
<td>Typical BWSp tumours (neuroblastoma, rhabdomyosarcoma, unilateral Wilms tumour, hepatoblastoma, adenocortical carcinoma or phaeochromocytoma)</td>
</tr>
<tr>
<td>Nephromegaly and/or hepatomegaly</td>
<td>Umbilical hernia and/or diastasis recti</td>
</tr>
<tr>
<td>Umbilical hernia and/or diastasis recti</td>
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</tbody>
</table>

**Notes**

1. For a clinical diagnosis of classical BWS a patient requires a score of 4 (this clinical diagnosis does not require the molecular confirmation of an 11p15 anomaly)
2. Patients with a score of ≥2 do not meet the criteria for genetic testing
3. Patients with a score of ≥2 with negative genetic testing should be considered for an alternative diagnosis
4. Patients with a score of ≥4 and this has been termed BWSp.

**BWS and hyperinsulinaemic hypoglycaemia**

The most common endocrine manifestation of BWS is hyperinsulinaemic hypoglycaemia (HH). This is a biochemical abnormality, characterised by the dysregulation of insulin secretion in the presence of low blood glucose levels. The biochemical diagnosis is made by demonstrating a detectable serum insulin (and/or C peptide) level with a low blood glucose level and low or suppressed fatty acids and ketone bodies. Approximately 50% of newborns with BWS will develop HH.

In the majority of cases of BWS, HH will be transient, lasting for a few days, and then resolve spontaneously. However, approximately 20% will have HH which persists beyond the first week of life, and about 5% will have severe HH which is medically unresponsive.5 The severe form of HH is observed in those children where the BWS is due to paternal UPD of chromosome 11p15. These cases traditionally require a near-total pancreatectomy.

**Recommendations for management**

The recent guidelines recommend that all neonates with suspected BWSp should be screened for HH before being discharged from hospital. HH which lasts less than 1 week is classified as a suggestive feature by the newly developed consensus criteria guidelines. In contrast, if the HH lasts beyond the first week of life, it is considered a cardinal feature. If a biochemical diagnosis of HH is confirmed, then the blood glucose levels should be maintained >3.9mmol/L in order to reduce the risk of brain damage.

In some patients with BWSp, the clinical features may be very subtle or may develop over time, and HH may be the only presenting biochemical feature.6 Thus, clinicians should have a low threshold for diagnosing BWSp.

The pathological basis of HH in patients with BWS is probably multifactorial and not completely understood. Possible mechanisms might include increased endocrine tissue mass, possible mutations in the gene ABC2/11/KCNJ11 and methylation defects.

The medical treatment options include diazoxide, short and long acting octreotide or mTOR inhibitors. Those patients who do not respond to medical therapy tend to improve when the mass of endocrine tissue is reduced by subtotal or near-total pancreatectomy.

**References**


Khalid Hussain

Professor of Pediatrics, Weill Cornell Medicine-Qatar, Division Chief – Endocrinology, Sidra Medicine, Doha, Qatar, and Honorary Professor, University College London UK
Leptin and lipodystrophy

Rebecca Brown and Marissa Lightbourne describe metreleptin’s role in management of lipodystrophy.

The discovery of leptin provided a mechanism to explain how the body regulates energy balance. Leptin deficiency, either in the physiologic state of starvation, or in pathophysiological states such as congenital leptin deficiency, causes hyperphagia, which is reversible with leptin replacement. However, pharmacologic doses of leptin do little to suppress appetite in states of endogenous leptin sufficiency or excess, such as obesity.

Leptin’s greatest success as a therapeutic agent (as the drug ‘metreleptin’) has been in patients with generalised lipodystrophy, who have leptin deficiency resulting from deficient adipose tissue.

Characteristics of lipodystrophy

Due to a lack of adipose tissue as a buffer for postprandial nutrient influx, lipodystrophy syndromes are complicated by ectopic lipid deposition, leading to severe insulin resistance, diabetes, dyslipidaemia, and non-alcoholic fatty liver disease (NAFLD). Treating metabolic complications of lipodystrophy with conventional medications is quite challenging, with many patients failing to achieve adequate control.

Management of lipodystrophy is reviewed in the 2016 practice guidelines. In brief, lifestyle modification, including diet and exercise, is essential. Oral hypoglycaemic agents, especially metformin, are commonly used for insulin resistance and diabetes. Thiazolidinediones improve glycated haemoglobin (HbA1c), insulin resistance, triglycerides and NAFLD in partial lipodystrophy. Insulin, often at high doses, remains an effective therapy for patients with uncontrolled diabetes. Statins, fibrates and fish oil are the main medications for dyslipidaemia.

The role of metreleptin

Metreleptin therapy in patients with generalised lipodystrophy reduces hyperphagia, improves insulin resistance and diabetes, and reduces serum and hepatic triglycerides. Improvements in blood glucose and triglycerides are seen as soon as 1 week, and are maintained over long term follow-up. In some cases, improvements in insulin resistance are quite dramatic, with resolution of diabetes even in patients who require high doses of insulin. In patients with partial lipodystrophy, who have preservation of some fat depots and hence less severe leptin deficiency, metreleptin leads to more variable metabolic improvements, with the greatest benefit seen in patients with lower endogenous leptin and more severe metabolic disease.

In children with generalised lipodystrophy, metreleptin reduced triglycerides by 50–60%. However, improvements were more noticeable in adolescents >12 years of age versus children ≤12 years, as adolescents had a greater baseline disease burden related to pubertal insulin resistance. Similarly, substantial reductions in HbA1c, alanine aminotransferase and aspartate aminotransferase were seen in adolescents, with smaller improvements in children. Metreleptin improved biopsy measures of NAFLD, reducing steatosis and ballooning injury, without changes in fibrosis. Overall, metabolic disease remained stable in children, with on-treatment levels comparable with those in adolescents. This suggests that metreleptin in children prevented worsening of metabolic disease during puberty that is part of the natural history of lipodystrophy.

Dosing and adverse effects

Metreleptin is approved in Japan and Europe for treatment of patients with both generalised and partial forms of lipodystrophy, and in the USA for generalised lipodystrophy only. Dosing in children <40kg is weight-based (starting dose ~0.06mg/kg per day). Although US and European labels for metreleptin provide fixed starting doses for patients >40kg (2.5mg/day in males, 5mg/day in females), most studies used weight-based dosing regardless of age or weight, with a maximum dose of 0.24mg/kg per day or 10mg/day, whichever is lower. Dose adjustments should be made based on normal growth in children, tolerability issues such as excessive weight loss, or inadequate clinical response. Patients with partial lipodystrophy often require higher doses of metreleptin versus more leptin-deficient patients with generalised lipodystrophy.

Key potential adverse effects of metreleptin include injection site reactions, weight loss (due to appetite suppression), hypoglycaemia (due to increased insulin sensitivity) in patients taking insulin or secretagogues, development of neutralising antibodies that may impair drug efficacy and potentially block endogenous leptin, and T cell lymphoma in patients with acquired generalised lipodystrophy.

Marissa Lightbourne
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Reference
ESPE: building bridges in rare diseases

This issue’s interview is with Faisal Ahmed and Rasa Verkauskiene, who answer our questions on ESPE’s current work in the field of rare diseases.

As we all know, the vast majority of endocrine diseases or conditions are rare. Recent initiatives, such as the European Reference Network on Rare Endocrine Conditions (Endo-ERN) and European Reference Network on Rare Bone Diseases (ERN BOND), have been welcomed as ways of increasing communication and knowledge-sharing among practitioners working on the diverse conditions that constitute the field of rare diseases.

In 2021, the ESPE Rare Disease Advisory Group (RDAG) began its work to review ESPE’s activities in this field. Its aim is to identify gaps and overlaps between the work of ESPE committees and other organisations, such as the ERNs and international patient organisations. The Group also engages with other societies, such as the European Society of Endocrinology (ESE).

We took this opportunity to ask Faisal Ahmed and Rasa Verkauskiene, who chair the RDAG, about the Group’s progress and what it means for the work of ESPE.

What is the RDAG set to achieve?

ESPE is heavily involved in supporting its members’ activities in the field of rare diseases. These may relate to education and training, clinical practice, research and advocacy. For instance, if you look at the curricula of ESPE training schools, the guidelines ESPE develops or the support ESPE provides to researchers through the Science Committee and research grants, you will see that the majority of the Society’s focus is on rare diseases.

However, by their nature, professional scientific societies also encourage the development of ‘silos’ that each focus on their own specialist area.

More recently, ESPE’s involvement in rare disease projects such as Endo-ERN, ERN BOND, the European Registries for Rare Endocrine Conditions (EuRRECa) and the European Registry for Rare Bone and Mineral Conditions (EuRR-Bone), together with the Society’s partnership with the European Society of Endocrinology (ESE) on these projects, has shown how organisations and interest groups can work together in pursuing a common vision.

The RDAG’s review of ESPE’s current activities in rare diseases will identify gaps and overlaps both between ESPE committees and with other organisations involved in this field.

What progress can you report to date?

It’s all very exciting! The RDAG has made a huge amount of valuable progress in our work with other organisations (see below). Importantly, we have also mapped the activities that ESPE already undertakes in rare diseases. In doing so, we have identified gaps where the Society could take a lead. For instance, we could host an online facility that allows laboratories to display the diagnostic tests that they can perform, which could be searchable by paediatric endocrinologists. This would help resource-restricted centres, as well as research groups whose work relies on cutting edge diagnostic technology, to identify and contact appropriate laboratories.

Which other organisations are you working with?

Over the first year, the RDAG has pushed forward with the development of the tripartite memorandum of understanding between Endo-ERN, ESPE and ESE. This will lead to a better definition of how the three organisations will work together in the field of rare diseases. The RDAG has not only facilitated ESPE’s link with Endo-ERN, but also with ERN-BOND, which previously did not have any official links to ESPE.

To strengthen the links with ESE, the RDAG has identified an ESE member from the ESE Rare Disease Committee to join the ESPE RDAG. This will lead to greater synergy in developing activities that are of interest to both societies. The RDAG has also been working with ESE and EuRRECa on developing ESPE’s response to the European Health Data Space consultation.

What developments will ESPE members see?

As well as initiatives we have already mentioned, we think the links with ERNs will also show societies how they can work with patients, perform surveillance activities and operate as networks that can improve patient care and increase opportunities for participating in research and clinical trials. Joint efforts in conducting common educational activities will avoid fragmentation and overlap.

What do you think the long term benefits will be?

Over the longer term, it is important that ESPE increases its profile in the field of rare diseases. It is not sufficient to engage in individual and specific fields within rare diseases; it is also important to showcase these activities and have a portal that allows communication and liaison with other stakeholders.

How can members of ESPE support the RDAG?

At the moment, the RDAG has a fixed life of 2 years, and is advising ESPE Council about how ESPE should continue and build up the RDAG’s work. We would like to hear from you with your thoughts and feedback.

And your message to ESPE members?

‘Alone we are rare. Together we are strong.’

*Strapline and registered trademark of the National Organization of Rare Diseases; www.rarediseases.org
EVENTS

ESPE NEWSLETTER / ISSUE 55 / SPRING 2022

ESPE 2022
Submit your abstracts by 19 April 2022 (23.59 BST)
www.eurospe.org/meetings/2022/espe-2022/abstracts
Early bird registration deadline 20 June 2022 (23.59 BST)

Hypothalamic dysfunction in childhood
ESPE Science Symposium 2022
Princess Máxima Center, Utrecht, The Netherlands, 7–8 October 2022
This 2-day event will discuss the aetiology of genetic and acquired hypothalamic dysfunction in childhood, its consequences and new ways of management to improve outcome.
We will also focus on building new networks and collaboration within ESPE and Europe, together with patient organisations and Endo-ERN.

Prizes and grants
Submit your abstract to ESPE 2022 for the chance to win one of the following awards:
• Henning Andersen Prizes
• Undergraduate Achievement Award
• ESPE President Poster Awards
• Travel Grants
www.eurospe.org/grants-awards/espe-meeting-grants

Fraudulent registration websites
Please be aware that fraudulent websites have been in operation, selling fake registration to ESPE 2022. The ESPE website (at www.eurospe.org or www.espe2022.org) is the only official website where you can register to attend the ESPE Meeting.

Personalised medicine in paediatric endocrinology
15–17 September 2022, Rome, Italy
The tremendous advancement in molecular biology has led to innovative approaches to many endocrine conditions. These have permitted more accurate diagnoses, tailored therapies and adequate genetic counselling. Whereas personalised medicine has been increasingly applied to diagnose and treat endocrine disorders in adults over the last decade, its implementation in paediatrics is just beginning.
Consequently, ESPE 2022 will focus on the application of personalised medicine to the child with endocrine disorders. The main discoveries in genetic/genomic research, as well as their present and future impact on the management of paediatric conditions, will be extensively covered in plenary lectures and symposia.
We are proud to host the 60th Annual Meeting of ESPE, and are excited that Rome will be the venue for the first in-person ESPE Meeting after the long interruption caused by the COVID-19 pandemic. We believe that ‘the eternal city’ is the ideal location to resume and create new friendships, and to enjoy networking and collaboration.
Stefano Cianfarani (President)
Mariacarolina Salerno (Vice-President)
on behalf of the Local Organising Committee of ESPE 2022

www.espe2022.org

Next ESPE Connect Webinar
Should we screen for type 1 diabetes in children?
17 March 2022, 16.00–17.30 (CET)
Convenor: Senthil Senniappan (UK)
Expert talks, with Q&A in the panel discussion
• Introduction – Francesco Chiarelli (Italy)
• The stages of type 1 diabetes development in children – Moshe Phillip (Israel)
• FOR routine diabetes screening in the general population – Tadej Battelino (Slovenia)
• AGAINST routine diabetes screening in the general population – Carla Greenbaum (USA)
• Panel Discussion
Free for members of ESPE or affiliated societies; non-members £25
Reserve your place at www.eurospe.org/education/webinar-series
**Future meetings**

See [www.eurospe.org/meetings](http://www.eurospe.org/meetings) for details of all future meetings

### 60th Annual ESPE Meeting
15–17 September 2022
Rome, Italy

### 11th International Meeting of Pediatric Endocrinology
4–7 March 2023
Buenos Aires, Argentina

### 61st Annual ESPE Meeting
21–23 September 2023
The Hague, The Netherlands

### 62nd Annual ESPE Meeting
November 2024
Liverpool, UK

### 63rd Annual ESPE Meeting
May 2025
Copenhagen, Denmark

### Other Events

- **ESPE Connect Webinar:** Screening for type 1 diabetes
  - 17 March 2022
  - Online

- **ESPE Maghreb School**
  - 30 May–1 June 2022
  - Online and at venues in Algeria, Morocco and Tunisia

- **ESPE Caucasus & Central Asia School**
  - 21–24 September 2022
  - Tbilisi, Georgia

- **ESPE Science Symposium:** Hypothalamic dysfunction
  - 7–8 October 2022
  - Utrecht, The Netherlands

- **ASPED-ESPE Endocrine Academy**
  - Details to be confirmed

### Deadlines

**APRIL**

- ESPE 2022 abstract submissions – 19 April 2022
- ESPE Research Unit final applications – 20 April 2022
- ESPE Research Fellowship applications – 20 April 2022

**MAY**

- ESPE Early Career Scientific Development Grant applications – 31 May 2022

**JUNE**

- ESPE 2022 early bird registration – 20 June 2022

For more information about vacancies on ESPE Committees and how to apply, see [www.eurospe.org/about/vacancies](http://www.eurospe.org/about/vacancies)

All dates, deadlines and plans are being constantly reviewed in light of COVID-19