INCONTINENTIA PIGMENTI (IP)  
(Bloch-Sulzberger syndrome)

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How common is IP?  
Incontinentia Pigmenti is a rare condition, affecting girls. Only in exceptional circumstances does it affect boys.

What are the features of IP?  
IP affects many parts of the body. It can affect different people to different degrees.

Skin features  
There are typically four stages that occur one after the other, though they may overlap. No specific treatment is needed for the skin changes.

Stage 1  
There is redness of the skin, then blistering, starting from the first few weeks of life. The blisters do not affect the face but occur in lines along the limbs and round the body. →

Blistering usually stops by four months of age, though blisters may recur at times when the child has a temperature. During this stage, the blisters should be kept clean and dry.

Stage 2  
As the blisters heal, warty areas occur on the skin of the hands and feet. In most cases these clear by six months of age.

Stage 3  
This is the stage that gives the condition its name. There are streaks and whorls of pigment along the limbs and round the body. These darken initially, then fade, usually by the age of 16 years.

Stage 4  
In adults there are pale, hairless streaks, best seen with an ultra violet light on the backs of the calves.

Nails  
About 40% of people with IP have fragile or ridged nails. Some have non-cancerous lumps under the nails that can be painful and can be removed surgically.

Hair  
Some girls with IP have thin hair and a few may have bald patches during the blistering stage. Later, half the people with IP have course, dull hair. Hair colour is normal.
Eyes
One third of girls with IP have a squint. They should be checked to see if they need glasses in order to prevent more serious visual problems.
More than 90% of people with IP have normal vision but some have a problem with the blood vessels in the back of the eye (retina). If present, this usually only causes a problem with one eye. Girls should have their eyes checked each month for the first few months. In some cases specialist treatment of the eye may be required.

Teeth
Over 80% of people with IP have late eruption of the teeth or fewer teeth than normal. Sometimes the teeth are a conical shape.

Breast
A small number of women may have some asymmetry in the size and shape of their breasts.

Development
Early studies showed that girls with IP often had problems with their development but more recent and more accurate studies have shown this not to be the case. Less than 10% of girls with IP have developmental or learning problems. Those who do have problems with development may have fits in the newborn period.

Genetics of IP
IP is caused by a change in a gene called NEMO. This gene is on the X chromosome. We have a total of 46 chromosomes in each cell of our body. Most of these are the same in both sexes but females have two X chromosomes and males have an X and a Y chromosome. If a male fetus had an altered NEMO gene it would almost certainly miscarry. Females can tolerate having one altered NEMO gene on one X chromosome as the other, normal gene on the other X chromosome, dilutes the effect of the altered gene.
In some cases the altered NEMO gene is passed from mother to daughter. In other cases the NEMO gene is normal in both parents but a change occurs in the gene when it is passed to the egg or sperm that made the girl.

Genetic testing
Since the recent discovery of the NEMO gene, it has been possible to test the gene. This may be done to confirm the diagnosis and to see whether the mother is affected. This may be important to establish whether IP could occur again in other pregnancies. Testing in pregnancy may be offered.

For more information about the genetics of IP
You can ask to be referred to your nearest Department of Clinical Genetics.

A Tribute to Richard
This is one tribute that I prayed I would never have to write, says Diana.
Richard had two very rare genetic conditions Hypogammaglobulineamia, a Primary Immunodeficiency and Hypohidrotic Ectodermal Dysplasia. He has battled for his life many times with diseases such as meningitis, bronchietasis, which severely damaged his lungs, whooping cough, pneumonia, Parvo Virus and collapsed lungs.
In the autumn 2001, after anxiously waiting for nearly two years for a donor, Richard underwent a lung transplant, which at the time was successful, but sadly in March Richard’s body began to reject them and he died on the 6th June 2002. Richard was almost 14.
Many of you will have read in EDlines Issue 7 September 1999 an article “Child of Achievement Awards” which told of the wonderful day Richard was invited to attend the Award Ceremony at the Park Lane Hilton, London to receive his Award. He was presented with a signed certificate and book of the event by Konnie Haq (Blue Peter) and Richard McCourt (Live and Kicking) which Richard was absolutely thrilled about and made his day. To win such an Award children will have undergone harrowing injuries or operations and in Richard’s case years of battling with life threatening illnesses.
Our thoughts and prayers go to his family and in particular, his parents David and Dayline.
Membership
We are still awaiting the return of around half the membership forms sent out. Incidentally, the New Members Pack was posted to all members, not emailed. If you didn't receive the Pack, please let us know, as it is important you receive all the information.

T.R.P. Syndrome
One of our members, Liz Greenfield, has written to inform us that she is the contact name through 'Contact A Family' for Trichorhinophalangeal Syndrome, and would be happy to talk to other members with this condition. Please contact the ED Society office for her details.

Tattoo
"Has anyone with ED experienced any problems with having a tattoo? Did they take any special measures." One of our members has a son with ED who is interested in having a tattoo on his upper arm, and would like to know how anyone else has got on. Please let us know if you have any information and can help.

Hair & Wigs
One of our members would like contact with anyone who has experience of their hair thinning over the years, to the extent that a wig becomes necessary. Would you like to chat with Audrey? Please contact us.

Terrific News!
One of our members has at last been given funding by his local council for air-conditioning in his home. This has taken a year to achieve and required the help of the Ectodermal Dysplasia Society, in particular Mandy White, one of our Trustees, and in the latter stages, the intervention of his local Member of Parliament. Due to the nature of the case, we are now in a position to confidently assist our other members in achieving this kind of help and, to this end, will write a more detailed article for the next Newsletter.

Feed-back on blistering
Two of our members had recently requested information on 'skin blistering', which we asked about in our April 2002 newsletter. We've had a response from an Italian member whose son experiences 'white flecks'. We are now interested to know whether members have noticed un-affected siblings being affected by these blisters as well, or is it limited only to those with ED? There is a medical view that blistering may not be specific to ED sufferers and may actually be common in those with other skin conditions. Please contact us.

Thanks for your help
A HUGE Thank You! A terrific amount has been donated, largely due to the membership drive. Specific donations have also been received, including from Southbroom Junior School; Legal & General Branch Mufti-Day; a Table-top Sale; Doctors' Conference Day and many of you have sent in stamps. Specific donations amounted to £176.95 - a tremendous effort. Thank you to all involved.

We were particularly impressed by Nadia (14), who used a Doctor's Conference to which she had been invited to attend as a patient, for an opportunity to raise money. Thankfully, the doctors paid up! Well done, Nadia!

Well done too to Oliver who also impressed us by running a cake stall at his School in his lunch break, to raise money for The ED Society.

Will this help?
Julia Errington who runs 'Face-the-Future', is one of our members and says that although she is allergic to many things, she has found the eye ointment S.S.P. (simple soft paraffin) has been particularly useful to her. Her doctor has prescribed this from Moorfields Eye Hospital as Julia is allergic to Lanolin and she has severe dry eye syndrome, corneal erosion and no tear ducts. Julia has asked us to pass this information on.

The use of a product name does not constitute a recommendation or endorsement by The Society.
Alison grew up believing she was born unlucky and would never find true happiness...

As Alison Smith talks to her husband, Nigel, one of their young children reaches up and tries to grab Alison's hair. Catching his hand she gently scolds him. Nothing out of the ordinary in that, except that she isn't worried about her hairstyle - just that what's underneath might be revealed. Despite being just 20 years old (21 in July) Alison wears a wig to hide baldness and ugly scars on her scalp.

She suffers from a rare condition called Ectodermal Dysplasia and has worn a wig, a hearing aid and dentures since she was a child. 'I don't mind so much now because I'm used to it, but as a child I hated being different to everyone else,' she says. 'I spent so much time in hospital that it became almost my second home.'

Although ED is a genetic condition neither of her parents had shown any sign of it and Alison wasn't diagnosed herself until she underwent an operation to correct a cleft palate when she was a toddler. As a baby, she'd suffered severe cradle cap and developed an infection on her scalp. The infection was scraped off and the hair on the top of her head never grew back. 'My GP sent me to have plastic surgery on my scalp,' says Alison. 'I had countless tissue expansion operations to try and make my hair grow back, but it never worked and my scalp was left covered in scars.'

By the time she started school Alison also had a scar behind her left ear after failed operations to fix her hearing impairment and her teeth were badly misshaped and had little enamel so they were removed.

'I didn't know I had ED because by the time my parents thought I was old enough to understand I had two younger sisters and a brother - all free from ED - and there was never a quiet moment to sit down and talk to me about it. I just thought I'd been born unlucky because I had so many health problems and looked different to everyone else.'

That changed when Alison started high school when, for the first time, she was teased and bullied by other pupils because of her appearance. The problem was made worse when at the age of 12, Alison turned up for school wearing her first NHS wig. 'The previous day I'd gone to school with my usual patchy, pale, brittle hair and scarred scalp and then the next day, I turned up with a full head of perfect hair,' she says. 'I was called 'wiggy' and the subject of everyone's jokes.'

When she was 14 Alison demanded that something was done about the bullying. Instead of addressing the bullying the school brought in a Counsellor to help Alison deal with it. It was the Counsellor who told Alison she was suffering from ED and put her in contact with the ED Support Group. 'It was fantastic knowing, at last, that I was different for a reason,' she says. 'I hadn't just been born unlucky. But knowing why I was different didn't stop the bullying.'

She spent much of her time writing poetry and stories often starring herself, but as a beautiful girl with long permed hair who grew up to marry and have children - something Alison was convinced could never happen to her.

'I believed my looks meant no one could ever fall in love with me and I'd spent so much time in hospital, I convinced myself my family would be better off without me,' says Alison.

At school, a few days before her 15th birthday Alison swallowed a packet of paracetamol tablets. Luckily a friend suspected something was wrong and searching Alison's bag discovered the
empty paracetamol packet and alerted their teacher. She was rushed to hospital where her stomach was pumped and the reaction from her family and friends proved to Alison how much she was loved and cared about.

After leaving school at 17 Alison started a one-year health and social care course at her local college in Norwich. She and her friends used to communicate with other students using an internal computer mailing network and one day Alison starting chatting online with a student called Nigel. After several computer chats and a quick glimpse of each other in a corridor Nigel asked Alison on a date.

I was really excited because despite having seen me he was still interested, but I was terrified of being rejected once he’d found out about ED,’ she says.

A few weeks later and Alison’s opinions on her future had changed radically. She says: ‘I was really falling for Nigel and he told me he felt the same way. For the first time I actually believed that it could be possible for me to have a normal relationship and even perhaps marriage.’

Three months after they’d begun dating Alison still hadn’t told Nigel about her condition and it was obvious to him that something was on her mind. He told her she shouldn’t be scared to tell him so she told him a little about her ED. Her rejection fears proved totally unfounded.

‘It didn’t bother me in the slightest. I liked Alison for who she was not whether she had her own hair,’ remembers Nigel, a supermarket canteen supervisor. ‘She’d never let me touch her hair so I’d had a few suspicions anyway.’

The following month Alison was shocked to discover she was pregnant. ‘I’d always assumed I wouldn’t be able to have children because of my health problems and although a Geneticist revealed our baby would have a 50 per cent chance of inheriting ED, we were overjoyed at the thought of becoming parents,’ she says.

The couple moved into a council flat together and while she was pregnant, Nigel once again proved his love and commitment to Alison when they met up after a dentist appointment. ‘I told Nigel I needed a new set and he asked ’a new set of what?’ He’d never realised I wore false teeth and it’d never occurred to me to tell him. But even my dentures didn’t faze him!’

She sailed through her pregnancy and Robert was born in December 1998 and was immediately diagnosed with ED. ‘I was a little anxious,’ says Alison. ‘My ED hadn’t bothered Nigel but now I’d given birth to his son who’d inherited my condition.

But again, it just wasn’t a problem for him.’

Nine months later Alison and Nigel, who were both 19, shared their perfect wedding and within months they began discussing the possibility of extending their family. Alison quickly fell pregnant again and at her first scan they were told they were expecting twins.

‘Having coped with ED myself I knew I’d be able to help Robert understand his condition and although we didn’t want the twins to have ED too, if they did then we could deal with it.’

Victoria and Alisha-Jane were born last November and were free of ED. Since then the family have moved into their own five-bedroom house, which they are renovating.

‘I’m still insecure about my looks and probably always will be,’ says Alison. ‘Nigel hasn’t seen me without my wig or a headscarf on. I know it wouldn’t change the way he feels about me, but I’m worried it might change the way I view our relationship.

‘Growing up, I just wanted to look normal because I thought only normal looking people could find happiness - but thanks to Nigel I’ve proved that it doesn’t matter what you look like - there’s love and happiness out there for everyone.’
FEED-BACK ON EUROPEAN CONFERENCE – SWEDEN 2002

The first European conference on the Ectodermal Dysplasias took place in Malmo, Sweden on the 30th-31st May 2002. The mission was to improve the care offered to European patients and to establish a network of professionals who want to develop oral health care services for individuals with ED in the European countries.

There were speakers from many countries giving presentations on subjects such as

- The role of patient organisations
- Epidemiology and clinical signs
- Oral signs and symptoms in ED
- Genetic counselling on ectodermal dysplasias
- Molecular background of defective hair and tooth development
- Approaching the young child with ED
- Oral and general care programmes
- Dental treatment of the young child with ED
- Prosthodontic treatment of the young child
- The link of professionals with patient organisations
- Analysis of mandibular growth after implants
- Oral rehabilitation in ED
- European efforts on centralisation of cleft lip and palate services

Whilst all these subjects were very interesting the main points which came out of this conference were that there is a great need to

- establish a formal network of centres and others working in the specialist area of the ectodermal dysplasias
- collaboration on merging databases both clinical and research to avoid duplication of effort (with appropriate safeguards to ensure data protection)
- work towards agreed protocols for diagnosis and multidisciplinary working as well as clinical guidelines for care that are patient driven
- collaboration on research projects in order to realise funding

The first step towards achieving the mission is to establish an Organising Team to create a database of contacts from the conference and generate a core working group to address the needs outlined during the conference in a rolling programme.

I feel very excited by the results of this conference and privileged that the ED Society has been invited to become involved from its conception. The patient organisations around the world, such as the Ectodermal Dysplasia Society will play an important role in bringing the needs of the people with ED to the professionals. We will be able to ensure that the professional focus is kept on the real people by keeping them informed of the work of our organisations and members.

I will continue to keep you informed of progress.

From the Trustees

Since the last Newsletter, the Executive Committee met on 18th May 2002. Subjects discussed were:

- Insurance – the Treasurer and Diana are still in liaison with the insurance company in relation to Public and Private Liability Insurance for EDS
- Fund-raising – the Chairman to produce framework + consider fundraising charity contract
- Balance Sheet – being updated and circulated each month to all Executive Committee members
- Membership – the Treasurer & Mary liaising re recording/processing membership fees
- Communication – David continues to keep the website up-to-date. There are now 14 medical/scientific articles on the website, plus 12 personal stories (real names omitted)
- New Members Pack – this has been mailed, and will be used for all future new members
• Press Pack – work continues on creating a press pack, and media guidelines for members
• Update – we have 178 children under 21 on our records, many of whom are not receiving the treatment to which they are entitled. Diana will emphasise the importance of early treatment at the ED Dental Conference in Sweden. We are liaising with a graphics art designer on the re-designing of the Charity’s logo, and also with a different printing company to see if we can get costs down even further on the production of newsletters and other mailings
• Expenses – a policy has been agreed and circulated to all Medical Advisory Board members for the reimbursement of travel expenses
• Air Conditioning – Mandy updated the committee – see ‘Terrific News’ in News & Views, p3.
• Family Conference 2003 – work has started on booking speakers and producing a programme

DENTAL IMPLANTS

One of our younger members, Gary Coburn, writes:

A few years ago in the year 2000 I underwent an operation where I received dental implants on my lower jaw. I had received surgery prior to that two years before when I had my upper jaw brought forward, as it was not growing at the same rate as my lower jaw and therefore the doctors told me I had to get an operation. This was very difficult for me as I am a bit of a coward when it comes to pain. However, I went through with both operations and I am able to eat with a stronger assurance that my lower denture will not move around or come out, which is a good feeling. Also I feel more confident about my looks, as before my operation I would get bullied at school with kids calling me ‘gremlin’ because of my uneven jaw line.

I would recommend the dental implants to anybody who is in need of them, as it has greatly helped me in my life in many different ways. I would be glad to answer any queries from anybody who is going to or is thinking of receiving this surgery, as I know it is a scary concept to go under the knife. I would like to say to anyone who may feel this way, what my Dad said to me when I didn’t want to go through with the two operations. My Dad said, “Gary, it is just a few weeks out of your life where you may feel a bit of pain; it’s worth it as you will enjoy the result for the rest of your life.”

The dental implants were in two stages where they implanted the foundations of the implant in my lower jaw bone and then a few months later I went back to the hospital and they put the second part of the implant in my mouth. Then I went to my dentist and he made me a new denture to sit on the implants, where now the implant holds my denture firmly in place, and I am really pleased with the results.

I would like to take this opportunity to wish anyone who is considering going through this operation all the best, and I hope you will be as glad as I am with the results.

Gary will be happy to chat to anyone regarding implants or how ED affects him. If you would like to talk to Gary, contact Diana at the ED Society office.

And still on the subject of teeth! ...

Stella Nicholas shares her daughter’s story:

My 4-year-old daughter, Joanna, is now 8 months into Reception Class at school and has been wearing dentures on a plate, which she calls her “brace”, for 3 months now.

At a recent social event, her school’s Family Fun Night, the children’s entertainer gave all the children clues of something he wished them to find amongst the grown-ups and take to him for a prize. The first idea was that they were supposed to get a comb that had “teeth” on it. Then they were to find something else with “teeth”. I will not forget the look on his face when Joanna promptly pulled out her “teeth” and slapped them into his hands. The joke was that most people thought that the “false teeth” they were trying to find belonged to an adult! Needless to say, she won the prize with lots of congratulations from her friends.

I wish to tell you this story as it sums up how proud I am of the way she has dealt with coping with her “brace”. Months ago when we realised it was the only way she could have ‘normal’ looking teeth as a child, I was very depressed. I worried about how she would cope with dealing with her teeth, starting school, and what other children would say.
Joanna is very open about her teeth. After lunch she always has a little group in the toilets with her watching her clean her teeth! Recently she had a new brace made that had more teeth put on. On her return to school the whole class gathered to admire the ‘new’ teeth.

She understands why she needs the brace and likes to tell people about her own teeth “not growing properly”.

Stella writes that she hopes this letter helps anybody else who may be worried about how their young child may cope with dentures.

And finally …

It has been a hectic first half of 2002 for me, we have a lot of new members joining us which is greatly encouraging and shows that the website is a very important tool in helping people find the Society. Thank you David for maintaining and updating the website.

Family Conference - 14th April 2003.

The registration paperwork for the Conference next year will be sent out with the October newsletter.

Many of you have still not returned your pink Membership Application Forms; could you please do this now. Don’t forget if you do not return your form it will be assumed that you no longer wish to be a member of the ED Society. Even if you have been a member since the Group started back in the 1980s; we still need your form. So please, return the completed pink form today.

As you will have read, we have been successful in helping one of our members obtain air-conditioning units from the Local Authority. This is wonderful news and shows, with our collective efforts, what can be achieved. Well done Mandy and thank you for all your hard work.

I am really pleased and grateful to our younger members who have taken the initiative to fundraise for the ED Society. I have realised that many people will sponsor our young for whatever activity they choose, whether it be a table-top sale, a 2-mile walk, a swimming event, baking cakes, etc. You don’t have to be sponsored for huge amounts: £5.00 goes a long way to paying the telephone bill!

So come on all you youngsters, summer’s coming, take Mum and Dad for a walk, cook Sunday lunch and Dad’s, how about a few rounds of golf or a sponsored short jog? Every little amount sent in helps us continue to help you. In fact, Ian Perry & Mary Rawson, two of our Trustees, are looking for sponsorship to walk part of the Pennine Way from 29 August to 2 September. Contact me if you want to sponsor them!

And now for some very exciting news! I was recently invited to give a presentation at the European Conference on Ectodermal Dysplasia in Sweden (see article on p6). The Conference was sponsored by the Swedish National Board of Health and Welfare and they paid for all my travel and accommodation expenses. The patient organisations from Austria, Sweden, Norway, Italy, USA and France were also invited to give a presentation.

I am very excited that the professionals are contemplating follow-up meetings, to give us the opportunity to bring more awareness of the patient support groups to professionals from all around the world. I feel for us (the members and Society) that this is a new beginning and there are many issues that need to be highlighted and addressed. It is important therefore, that you all share your concerns and problems with me so that I can relay these to the professionals to obtain answers for each and every one of us.

I hope you all have a very enjoyable summer.

Diana

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**Trustees (Executive Committee Members)**

- Chairman: Paul Collacott
- Treasurer: Andrew Ponting
- Secretary: Mary Rawson
- Webmaster: David Wyatt
- Jean Miles
- John Moss
- Diana Perry
- Ian Perry
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**Medical Advisory Board Members and their fields**

- **Chairman**: Paul Collacott
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- **Paediatric Dental Surgery**: Peter Crawford
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- **Ophthalmology**: Stephen Kaye
- **Genetics, Molecular Dermatology**: John McGrath
- **Ear, Nose & Throat**: Richard Mills
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- **Paediatric Dermatology**: Celia Moss
- **Clinical Genetics (Incontinentia Pigmenti)**: June Nunn
- **Ophthalmology**: Helen Stewart
- **Clinical Genetics**: Colin Willoughby

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