proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).

proteinspotlight
> ONE MONTH, ONE PROTEIN <

Issue 136, March 2012

www.proteinspotlight.org

the ends of our fingers

Vivienne Baillie Gerritsen

Fingertips are hugely sensitive. And, besides the fairly recent mobile phones that rely on them entirely, we put their sensitivity to use constantly. They are able to grasp subtle differences in temperature and texture – such as discern warm from tepid for example, or a dry surface from a greasy one. They are also able to touch or feel extremely delicately. In truth, the ends of our fingers are able to give a pretty clear picture of what is happening around us. This, of course, is thanks to nerve ends which reach the very tips of them. But scientists are now suggesting that our digital refinement may also be partly due to the epidermal ridges which cover them. In other words: our fingerprints. Fingerprint architecture is slowly being uncovered, thanks to diseases that have the power to wipe them away. One such disease – known as adermatoglyphia – is caused by a deficiency in a protein known as SMARCAD1.

Andreas Roseneder, *thumbnails & fingertips*

Chinese ink & AquaBrique on Kahari-paper 2011

Courtesy of the artist

In 1684, long before scientists delved into the molecular origins of our fingerprints, a British doctor known as Nehemiah Grew gave a lecture on the patterns formed by the ridges on the ends of human fingers. It was the very beginning of an awareness of the uniqueness of fingerprints. Very little interest, however, was given to the subject for the best part of two centuries until William James Herschel - an employee of the East India Company faced with high rates of local illiteracy – started to use handprints to seal contracts before turning to fingerprints as a personal signature. From then on, and due to a combination of circumstances, the use of fingerprints as a means of identification was gradually accepted and by the beginning of the 20th century, they were used not only to catalogue criminals but also to identify them.

Unsurprisingly, the power of fingerprints has spread to customs and passports. We now know that the patterns formed by the epidermal ridges on our fingertips are unique – the same pattern shared only by identical twins – and are still the surest way of identifying someone. Providing they have any… Indeed, there are very rare cases in which individuals present no fingerprints at all, because of an absence of epidermal ridges. This can be due to diseases that cause other severe handicaps, but there is also a case of adermatoglyphia which has no particular incidence on the individual other than smooth ends of fingers and problems at immigration – which is the reason the condition has also been baptised “immigration delay disease”. The first occurrence of such a disease arose from the case report of a young Swiss woman who had recurring trouble trying to get through USA checkpoints that use computerized fingerprint recognition.

There are life-circumstances in which people can lose their fingerprints: accidents, swelling,

self-infliction and various types of labour for example. But none of this applied to the young Swiss woman. Scientists began to take an interest in her and discovered that her fingers, like her palms, toes and soles were devoid of epidermal ridges. What is more, certain members of her family presented the same problem. So the condition was hereditary. And besides a reduced amount of sweat glands, there was no other physiological complaint. So they started a search for the gene which could be at the heart of it all. They discovered an isoform that was horrendously-baptised "SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1" (or SMARCAD1…), but is otherwise known as ATP-dependent helicase 1 or hHEL1.

hHEL1 is expected to be small and globular in structure and is only expressed in the skin – which made it an ideal candidate for adermatoglyphia. It belongs to the helicase superfamily – enzymes that are able to unwind DNA or RNA, and are hence involved in gene expression and protein production. What is more, hHEL1 also seems to be able to interact not only with nucleic acids but also with other proteins. So what was the Swiss woman's hHel1? Well, a mutation within the hHEL1 gene was found to be common to all the family members suffering from adermatoglyphia, and is believed to wipe out its function altogether. But how can it obliterate fingerprints?

As always, the malfunction of a protein gives hindsight into what its natural function could be. Human fingerprints are forged during the embryo's life in utero, and are believed to have both a genetic and environmental origin. Pads under the growing embryo's skin, known as volar pads, gradually disappear whilst simultaneously forming the epidermal ridges on our fingertips. By the sixth month, a foetus's fingerprints are the ones it will have during its lifetime. It so happens that the sweat glands also form along these ridges – which would explain the sweat deficiencies on the Swiss family's hands and feet. hHel1 must have a role in epidermal ridge architecture, and hence sweat gland formation. Perhaps via cell to cell contacts during development? So far, no one knows.

A question arises: what are fingerprints for in the first place? Surely not for promoting delays at immigration checkpoints… Until recently, it was thought that epidermal ridges were a way of increasing friction and thus giving hands, or indeed feet, a better grip. This theory seems to have lost its lustre however, and scientists now believe that the whole point of ridges may be a way of increasing sensitivity to finer sensations. From an evolutionary point of view, such an argument makes sense. Finer sensitivity spells finer tuning to an organism's environment, which is important for its survival. In the not so distant future, DNA may well replace fingerprints for human identification. This said, mutations such as those which cause adermatoglyphia are crucial in understanding how humans are made before they appear on this side of the world.

Cross-references to UniProt

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A containing DEAD/H box 1, *Homo sapiens* (Human) : Q9H4L7

References

1. Nousbeck J., Burger B., Fuchs-Telem D., Pavlovsky M., Fenig S., Sarig O., Itin P., Sprecher E.
   A mutation in a skin-specific isoform of SMARCAD1 causes autosomal-dominant adermatoglyphia
   The American Journal of Human Genetics 89:302-307(2011)
   PMID: 21820097

2. Burger B., Fuchs D., Sprecher E., Itin P.
   The immigration delay disease: Adermatoglyphia-inherited absence of epidermal ridges
   Journal of the American Academy of Dermatology 64:974-980(2011)
   PMID: 20619487

Protein Spotlight (ISSN 1424-4721), http://www.proteinspotlight.org, is published by the Swiss-Prot group at the Swiss Institute of Bioinformatics (SIB).