

UCL INSTITUTE OF CHILD HEALTH

DEVELOPMENTAL ENDOCRINOLOGY RESEARCH GROUP
CLINICAL & MOLECULAR GENETICS UNIT

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PARENT/GUARDIAN INFORMATION SHEET

Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)

Name of Principal Investigator: Dr John Achermann

Dear

You and your child are being invited to take part in a research study into *Novel Mechanisms in Reproductive Biology*.

This project is based at UCL Institute of Child Health and is supported by The Wellcome Trust and/or European Union.

Before you decide whether to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully. Talk to others if you wish.

- Part 1 of this information sheet tells you the purpose of this study and what will happen if you take part
- Part 2 of this information sheet gives you more detailed information about the conduct of this study

Please feel free to ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

We thank you for taking the time to read this.

Sincerely

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Hospital NHS Trust

What is the aim of the study?

The aim of this study is to find new mechanisms involved in the development and function of the reproductive system (gonad development, puberty). We hope to find new genes and proteins involved in reproductive biology, and to work out how variations in known genes and proteins have their effect.

Why has my child been chosen?

We are approaching individuals attending our clinic.

Why is this study being done?

Within the past twenty years, we have started to understand some of the processes that control reproductive development and function in humans.

These processes are controlled by **proteins**, such as growth factors in the testis or ovary, or hormones in the blood. These proteins are encoded by **genes**, which are regions of each person's DNA blueprint or genetic code found on the **chromosomes**.

By analysing the proteins/hormones, genes and chromosomes of people with disorders of reproductive development in more detail we hope to find new factors involved in reproductive biology. We also want to improve our knowledge of those factors we already know are important, such as steroid hormone enzymes and receptors.

The results of our research will help us to understand reproductive development and function better. These findings could help us to improve the diagnosis and management of individuals and families with reproductive system disorders.

Does my child have to take part?

No. It is up to you whether or not your child takes part. If they do, you will be given this information sheet to keep and be asked to sign a consent form. Your child is still free to withdraw at any time and without giving a reason. A decision to withdraw at any time, or a decision not to take part, will not affect the standard of care your child receives.

What will happen to my child if we are interested in taking part?

If you and your child agree to participate in the study, we would obtain a small blood sample (one to two teaspoonfuls depending on age) or saliva sample (two teaspoonfuls, using a special kit, in children older than 6 years) to extract DNA. This DNA will be stored and analysed for changes in specific genes related to reproductive biology. Some of these genes are currently known; others we hope to identify in the future. Wherever possible, we will take these blood samples at the same time that routine clinical tests are being done.

We might ask that your child provides an additional blood sample (one to two teaspoonfuls depending on age) to allow us to analyse hormones and proteins present in the blood stream, and to grow blood cells to perform more detailed analysis of chromosomes. We would be looking for small changes in chromosome structure that may not be picked up on routine clinical tests. Some of these detailed chromosome analysis studies can also be performed on DNA.

Usually only single samples are requested, although we may request an extra sample from your child if we find something we would like to investigate in more detail, and we may ask for additional blood samples to look at the hormone or protein pattern.

We also request your consent to take urine samples for hormone profiles in some situations, and to have your approval to retain some surplus pieces of the gonad (testis or ovary) or skin at the time of an operation if this ever occurred to allow us to perform more detailed analysis of genes, proteins and hormones in these tissues.

With your consent, we plan to keep the samples for this current study and for other relevant studies in the future, if there is enough sample left and if we receive funding for such projects.

Your participation in this study will not require additional clinic visits, although if you would like us to tell you any of our findings this may require slightly longer outpatients appointments, or a special appointment

arranged with myself or one of our clinical geneticists.

If you are happy to take part, and are satisfied with the explanations from your research team, you will be asked to sign a consent form. If your child is able to understand the research and is happy to take part and can write their name, they will be asked to sign an “assent” form with you, if they want to. You will be given a copy of the signed information sheet and consent/assent forms to keep for your records.

What are the alternatives for diagnosis or treatment?

The research tests proposed in this study are performed *in addition* to any standard clinical tests or investigations considered necessary as part of your child’s routine management. No routine tests or treatment would be withheld on the basis of whether you and your child decide to participate in this study or not.

What are the potential risks and disadvantages of taking part?

No significant physical risk can be foreseen. We will try to take any blood samples at the same time that routine clinical tests are being performed, if possible. If a blood sample is being taken specifically for this study there will be some discomfort from the needle prick, but local anaesthetic cream will be used to numb the skin first. If at any time you or your child feel that the actual or perceived distress is too great, please don’t hesitate to tell your research investigator. Issues related to genetic testing are discussed in more detail in Part 2.

What are the possible benefits of taking part?

There may be no immediate benefit to your child from taking part in this study, but our research will help us to understand reproductive development and function better. This information may help others with conditions affecting the reproductive system or puberty.

Also, there is a small possibility that we might find out more information related to your child’s specific condition. You will have the option whether any potential results are given to you. You can choose whether we give you an aggregate result (for example, 3 out of 20 participants had interesting findings) or whether we give you your actual results.

As this is a research study rather than an approved clinical “diagnostic” test, we would first check our findings on extra samples, and then repeat these studies in a clinical laboratory that undertakes genetic tests, if this is possible. The importance or implications of our research findings may not be known for some time, or clinical tests may not be readily available. It is very important to realize that our results are research findings and are not a clinical test. Further information related to genetic tests is provided in Part 2.

What happens when the research study stops?

We would like to keep any samples and relevant information related to this study for future research analysis beyond this current project. Undertaking further research work will be dependent on future funding.

What will happen if I or my child don’t want to carry on with the study?

You and your child’s participation in this study is voluntary. If you decide now or at a later stage that you do not wish to participate in this research project you are free to withdraw at any time, without giving any reason. Your child’s medical treatment or legal rights will not be affected. Stored samples will be destroyed if you wish.

What if there is a problem?

Any complaint about the way you or your child have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

Will my child’s taking part in the study be kept confidential?

Yes. All information about your participation will be kept confidential. The details are included in Part 2.

Contact details:

For further information at any stage, you are welcome to contact: Dr John Achermann, Wellcome Trust Senior Research Fellow in Clinical Science, Endocrinology, UCL Institute of Child Health, 30 Guilford Street, London WC1N 1EH (0207 905 2887 or j.achermann@ich.ucl.ac.uk).

This completes Part 1 of the Information Sheet. If you and your child are interested in participating, please continue to read the additional information in Part 2 before making any decision.

PART 2 – Detailed information

What if relevant new information becomes available?

Your participation in this research study will not interfere with your child being offered any relevant new tests or management approaches that might become available in the future.

Will my child's taking part in this study be kept confidential?

All information that is collected about your child during the course of the research will be kept strictly confidential and in compliance with the Data Protection Act 1998. Any paper records will be stored securely and any electronic records bearing "identifiers" (e.g., names) will be password protected. Samples in the Research Laboratory will be given a research number, which will be used during analysis rather than names.

Only the researchers and a representative of the Research Ethics Committee will have access to the data collected during the study. You will be asked to agree that appropriate sections of your child's medical notes may be looked at by responsible individuals from the Institute of Child Health where it is relevant to this research.

Notification of your General Practitioner (GP) and specialists involved in your child's care

We will ask you for your permission to inform your GP and other specialists about your child's participation in this study. In some situations it will be important to discuss potentially significant research findings with other members of the clinical and research team, and other specialists involved in your care.

What will happen to any samples my child gives?

Any samples provided will be processed initially in the Clinical Genetics Laboratory of Great Ormond Street Hospital (DNA extraction), Regional Cytogenetics Laboratory (chromosomes), or in the Research Laboratory at the Institute of Child Health (salivary DNA, serum/plasma) before being transferred for storage and analysis by the research team involved in this study at the Institute of Child Health.

Everyone taking part in this study will be given a study number ("coded") and samples kept in the research laboratory will be handled on a day-to-day basis using this number. Only the research team involved in this study will have access to these samples, and only the principal investigator and designated individuals will be able to identify whose sample it is.

We like to view any samples provided as "gifts" for the purposes of research. It is hoped that the samples will be kept for analysis by the research team on an ongoing basis, but it is quite possible that the samples will be used up during the course of this study. We also ask your permission to distribute coded samples and anonymous information to collaborators involved in related research within the UK, within the European Economic Area, and to countries outside of Europe. No names will be provided and you are free to choose whether you agree to this or not. With your consent, we may use some of the samples for commercial collaborations (i.e. partnerships between University and Industry) to develop better diagnostic and therapeutic tools. Commercial collaborators would not receive any identifying information, thus your personal information remains safe and confidential with the research team.

We do not anticipate that this work will lead to any outcome of commercial significance and you or your child would not benefit financially if this research does lead to the development of a new treatment or medical test.

Will any genetic tests be done?

The aim of this study is to identify new genes involved in reproductive development and function, or variations in known genes involved in these systems.

We will be able to tell you some of the genes we plan to look at when you start the study. We will also ask you whether we can analyse additional factors that may be relevant in reproductive biology as they are identified. You may allow us to do this without contacting you further, or you may request we contact you to get you and your child's agreement each time.

Although this is a research study, we are willing to share any potential positive findings with you. We may ask a clinical geneticist to join us to explain any findings. If you would prefer not to know the results of any of our investigations then that is your choice. **It is very important to appreciate that our results are research**

findings rather than clinical tests. We recommend checking any positive findings in a separate sample, and – if you and your child wish – trying to confirm our findings in a clinically approved laboratory. However, it is quite possible that such clinical tests are not readily available at the present time. It is also possible that the significance of our findings is unclear at present. Finally, it is not currently possible to analyse all genes and proteins in their entirety, so we might overlook subtle changes in our studies.

In a few instances, we are also trying to analyse many genes at once using a specially designed “gene chip”. This new technology might be useful in the future for obtaining more rapid results for patients. If we are interested in including your DNA in our study to test this new technology we will discuss it with you.

What will happen to the results of the research study?

Our aim is that the results of these studies will be presented to the scientific community and important advances will be published in peer-reviewed scientific and medical journals. This is the best way of having our work reviewed by experts in the field, and allowing other doctors and patients to benefit from our findings.

You and your child will not be identified in any report or publication unless you have consented to the release of such information. Clinical data, biochemical data (e.g. hormone results) and radiological (e.g. X-rays, scans), histological or whole-organ images (e.g. pictures of the gonad tissue) may be used. External photographs will not be used without specific consent.

What if there is a problem?

If you or your child have a concern about any aspect of this study please discuss them in the first instance with the principal researcher (Dr John Achermann, 0207 905 2887) who will do their best to answer your questions. If the problems are not resolved, or you wish to comment in any other way, please contact the Research and Development Office, Institute of Child Health, 30 Guilford Street, London WC1N 1EH (Emma Pendleton, 020 7905 2179).

This project has been approved by an independent Research Ethics Committee who believe that it is of minimal risk to you. However, research can carry unforeseen risks and we want you to be informed of your rights in the unlikely event that any harm should occur as a result of taking part in this study.

This research is covered by a no-fault compensation scheme, which may apply in the event of any significant harm resulting to you from involvement in the study. Under this scheme it would not be necessary for you to prove fault. You also have the right to claim damages in a court of law. This would require that you prove fault on the Hospital/Institute and/or any manufacturer involved.

Who is organizing and funding the research?

This study is being organized by Dr John Achermann at the UCL Institute of Child Health and is being funded by The Wellcome Trust as part of a Senior Research Fellowship in Clinical Science as well as by the European Union.

Who has reviewed the study?

This study has been reviewed and approved by the Research Ethics Committees of UCL Institute of Child Health/Great Ormond Street Hospital NHS Trust (REC reference 05/Q0508/24; Study number 05BC13).

If you and your child are interested in taking part you will be given a copy of this information sheet and any signed consent form to keep.

Please feel free to contact us if you have any further questions, or if your care moves to a different hospital or clinic.

We thank you for considering taking part and for taking the time to read this information sheet.

Contact:

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology
UCL Institute of Child Health
30 Guilford Street, London WC1N 1EH
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PARTICIPANT INFORMATION SHEET (14 YEARS +)

**Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)**

Name of Principal Investigator: Dr John Achermann

Dear

You are being invited to take part in a research study into *Novel Mechanisms in Reproductive Biology*.

This project is based at UCL Institute of Child Health and is supported by The Wellcome Trust and/or European Union.

Before you decide whether to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully. Talk to others if you wish.

- Part 1 of this information sheet tells you the purpose of this study and what will happen if you take part
- Part 2 of this information sheet gives you more detailed information about the conduct of this study

Please feel free to ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

We thank you for taking the time to read this.

Sincerely

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Hospital NHS Trust

What is the aim of the study?

The aim of this study is to find new mechanisms involved in the development and function of the reproductive system (puberty, testis or ovary development).

Why have I been chosen?

We are approaching young people attending our clinic for assessment of puberty.

Why is this study being done?

Within the past twenty years, we have started to understand some of the processes that control how the reproductive system works.

These processes are controlled by **proteins**, such as hormones in the blood. These proteins are made by **genes**, which are regions of each person's DNA blueprint or genetic code found on the **chromosomes**.

By analysing the proteins/hormones, genes and chromosomes of people with reproductive/puberty disorders in more detail we hope to find new factors involved in reproductive biology. We also want to improve our knowledge of those factors we already know are important, such as steroid hormone enzymes and receptors.

The results of our research will help us to understand reproductive development and puberty better. These findings could help us to improve the diagnosis and management of people with these conditions.

Do I have to take part?

No. It is up to you whether or not to take part. If you do, you will be given this information sheet to keep and be asked to sign a consent form. You are still free to withdraw at any time and without giving a reason. If you decide not to take part or to withdraw at any time it will not affect the care you receive from the Doctors or Hospital.

What will happen to me if I am interested in taking part?

If you agree to participate in the study, we will ask you to provide a small blood sample (two teaspoonfuls) or saliva sample (two teaspoonfuls, using a special kit) to extract DNA. This DNA will be stored and analysed for changes in specific genes related to reproductive biology. Some of these genes are currently known; others we hope to identify in the future. Wherever possible, we will take these blood samples at the same time that routine clinical tests are being done.

We might also ask you to provide additional blood samples (four teaspoonfuls, 20mls) to allow us to analyse hormones and proteins present in the blood stream, and to grow blood cells to look at the chromosomes in more detail. We would be looking for small changes in chromosome structure that may not be picked up on routine tests you may have had done already. Some of these detailed chromosome analysis studies can also be performed on DNA.

Usually only single samples are requested, although we may request an extra sample if we find something we would like to investigate in more detail.

We may also request your consent to take urine samples for hormone profiles in some situations.

If you ever have an operation that means some reproductive tissue (testis, ovary) is removed, we may ask you specifically if we can keep some of this for a more detailed analysis of genes, proteins and hormones.

With your consent, we plan to keep the samples for this current study and for other relevant studies in the future, if there is enough sample left and if we receive funding for such projects.

Your participation in this study will not require additional clinic visits, although if you would like us to tell you any of our findings this may require slightly longer outpatients appointments, or a special appointment arranged with myself or one of our clinical geneticists.

What are the alternatives for diagnosis or treatment?

The research tests proposed in this study are performed *in addition* to any standard clinical tests or investigations considered necessary as part of your routine clinical management. No routine tests or treatment would be withheld on the basis of whether you decide to participate in this study or not.

What are the potential risks and disadvantages of taking part?

We do not expect there to be any risks if you take part. We will try to take any blood samples at the same time that routine clinical tests are being performed, if possible. If a blood sample is being taken specifically for this study there will be some discomfort from the needle prick, but local anaesthetic cream will be used to numb the skin first.

Issues related to genetic testing are discussed in more detail in Part 2.

What are the possible benefits of taking part?

There may be no immediate benefit to you from taking part in this study, but our research will help us to understand reproductive development and function better. This information may help other people with conditions affecting puberty.

Also, there is a small possibility that we might find out more information related to your specific condition. You will have the option whether you are told any results of our research tests. You can choose whether we give you an aggregate result (for example, 3 out of 20 participants had interesting findings) or whether we give you your actual results.

If we do find something interesting in analysis of genes, we will ask for an extra blood sample to check our findings. We will also try to repeat our findings in a clinical laboratory that undertakes genetic tests, if you would like and if this is possible. Sometimes, our findings might need more detailed studies in the laboratory to know if they are important or not. It is very important to realize that our results are research findings and are not a clinical test.

What happens when the research study stops?

We would like to keep any samples and relevant information related to this study for future research analysis beyond this current project. Undertaking further research work will be dependent on future funding.

What will happen if I don't want to carry on with the study?

Your participation in this study is voluntary. If you decide now or at a later stage that you do not wish to participate in this research project you are free to withdraw at any time, without giving any reason. Your medical treatment or legal rights will not be affected. Any stored samples will be destroyed if you wish.

What if there is a problem?

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

Will my taking part in the study be kept confidential?

Yes. All information about your participation will be kept confidential. The details are included in Part 2.

Contact details:

For further information at any stage, you are welcome to contact: Dr John Achermann, Wellcome Trust Senior Research Fellow in Clinical Science, Endocrinology, UCL Institute of Child Health, 30 Guilford Street, London WC1N 1EH (0207 905 2887 or j.achermann@ich.ucl.ac.uk).

This completes Part 1 of the Information Sheet. If you are interested in participating, please continue to read the additional information in Part 2 before making any decision.

PART 2 – Detailed information

What if relevant new information becomes available?

Your participation in this research study will not interfere with you being offered any relevant new tests or management that might become available in the future.

Will my taking part in this study be kept confidential?

All information that is collected about you during the course of the research will be kept strictly confidential and in compliance with the Data Protection Act 1998. Any paper records will be stored securely and any electronic records bearing “identifiers” (e.g., names) will be password protected. Samples in the Research Laboratory will be given a research number, which will be used during analysis rather than names.

Only the researchers and a representative of the Research Ethics Committee will have access to the data collected during the study. You will be asked to agree that appropriate sections of your medical notes may be looked at by responsible individuals from the Institute of Child Health where it is relevant to this research.

Notification of your General Practitioner/Family Doctor (GP) and specialists involved in your care

We will ask you for your permission to inform your GP and other specialists about your taking part in this study. In some situations it will be important to discuss potentially significant research findings with other members of the clinical and research team, and other specialists involved in your care.

What will happen to any samples I give?

Any samples provided will be processed initially in the Clinical Genetics Laboratory of Great Ormond Street Hospital (DNA extraction), Regional Cytogenetics Laboratory (chromosomes), or in the Research Laboratory at the Institute of Child Health (salivary DNA, serum/plasma) before being transferred for storage and analysis by the research team involved in this study at the Institute of Child Health.

Everyone taking part in this study will be given a study number (“coded”) and samples kept in the research laboratory will be handled on a day-to-day basis using this number. Only the research team involved in this study will have access to these samples, and only the principal investigator and designated individuals will be able to identify whose sample it is.

We like to view any samples provided as “gifts” for the purposes of research. It is hoped that the samples will be kept for analysis by the research team on an ongoing basis, but it is quite possible that the samples will be used up during the course of this study. We also ask your permission to distribute coded samples and anonymous information to collaborators involved in related research within the UK, within the European Economic Area, and to countries outside of Europe. No names will be provided and you are free to choose whether you agree to this or not. With your consent, we may use some of the samples for commercial collaborations (i.e. partnerships between University and Industry) to develop better diagnostic and therapeutic tools. Commercial collaborators would not receive any identifying information, thus your personal information remains safe and confidential with the research team.

We do not anticipate that this work will lead to any outcome of commercial significance and you would not benefit financially if this research does lead to the development of a new treatment or medical test.

Will any genetic tests be done?

The aim of this study is to identify new genes involved in reproductive development and function, or variations in known genes involved in these systems.

We will be able to tell you some of the genes we plan to look at when you start the study. We will also ask you whether we can look at additional factors that may be relevant in reproductive biology as they are identified. You may allow us to do this without contacting you further, or you may request we contact you to get your agreement each time.

Although this is a research study, we are willing to share any potential positive findings with you. We may ask a clinical geneticist to join us to explain any findings. If you would prefer not to know the results of any of our investigations then that is your choice. **It is very important to appreciate that our results are research**

findings rather than clinical tests. We recommend checking any positive findings in a separate sample, and – if you wish – trying to confirm our findings in a clinically approved laboratory. However, it is quite possible that such clinical tests are not readily available at the present time. It is also possible that the significance of our findings is unclear at present. Finally, it is not currently possible to analyse all genes and proteins in their entirety, so we might miss small changes in our studies.

In a few instances, we are also trying to analyse many genes at once using a specially designed “gene chip”. This new technology might be useful in the future for obtaining more rapid results for patients. If we are interested in including your DNA in our study to test this new technology we will discuss it with you.

What will happen to the results of the research study?

Our aim is that the results of these studies will be presented to the scientific community and conferences and meeting and important advances will be published in peer-reviewed scientific and medical journals. This is the best way of having our work reviewed by experts in the field, and allowing other doctors and patients to benefit from our findings.

You will not be identified in any report or publication unless you have consented to the release of such information. Clinical data, biochemical data (e.g. hormone results) and radiological (e.g. X-rays, scans), histological or whole-organ images may be used. External photographs will not be used without asking your specific approval.

What if there is a problem?

If you are worried about any aspect of this study please discuss this in the first instance with the principal researcher (Dr John Achermann, 0207 905 2887) who will do their best to answer your questions. If the problems are not resolved, or you wish to comment in any other way, please contact the Research and Development Office, Institute of Child Health, 30 Guilford Street, London WC1N 1EH (Emma Pendleton, 020 7905 2179).

This project has been approved by an independent Research Ethics Committee who believe that it is of minimal risk to you. However, research can carry unforeseen risks and we want you to be informed of your rights in the unlikely event that any harm should occur as a result of taking part in this study.

This research is covered by a no-fault compensation scheme, which may apply in the event of any significant harm resulting to you from involvement in the study. Under this scheme it would not be necessary for you to prove fault. You also have the right to claim damages in a court of law. This would require that you prove fault on the Hospital/Institute and/or any manufacturer involved.

Who is organizing and funding the research?

This study is being organized by Dr John Achermann at the UCL Institute of Child Health and is being funded by The Wellcome Trust as part of a Senior Fellowship in Clinical Science as well as by the European Union.

Who has reviewed the study?

This study has been reviewed and approved by the Research Ethics Committees of UCL Institute of Child Health/Great Ormond Street Hospital NHS Trust (REC reference 05/Q0508/24; Study number 05BC13).

If you are interested in taking part you will be given a copy of this information sheet and any signed consent form to keep.

Please feel free to contact us if you have any further questions, or if your care moves to a different hospital or clinic.

We thank you for considering taking part and for taking the time to read this information sheet.

Contact:

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology
UCL Institute of Child Health
30 Guilford Street, London WC1N 1EH
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INFORMATION SHEET FOR CHILDREN UNDER 8 YEARS

Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)

Name of Principal Investigator: Dr John Achermann

Dear

We are doing a project at Great Ormond Street Hospital looking at the way parts of our bodies grow and develop.

We are asking some of the children who come to the clinic to help us with our work.

We would like to take an extra blood sample (one or two teaspoons) to look at some of the building blocks that make up our bodies. We may also ask for samples of urine (wee).

We will discuss this with your parents.

We would be very happy to answer any questions you have too.

It is up to you whether you want to do this.

You do not have to do this if you do not want to.

Thank you for reading and listening to this.

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Hospital NHS Trust

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INFORMATION SHEET FOR CHILDREN 8-14 YEARS

Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)

Name of Principal Investigator: Dr John Achermann

Dear

We are doing a project at Great Ormond Street Hospital looking at the way various parts of our bodies develop and work. We are interested in looking at the building blocks called genes and proteins that make up our bodies.

We are asking some of the children and young people who come to the clinic to help us with our work.

We would like to ask you if we can take an extra blood sample (one or two teaspoons) to look at some of the building blocks (genes and proteins) that make up our bodies. If you agree, we would try to take this sample at a time when you are having blood tests done anyway. We will use cream to make your skin numb before any samples are taken. Sometimes we will ask for a saliva (spit) sample instead of blood, or samples of urine (wee). We would like to keep these samples to use in our project and future projects.

We will discuss this with your parents and ask for their approval too.

We would be very happy to answer any questions you have. We would be very happy to explain this work in more detail if you would like us to.

It is up to you whether you want to take part.

You do not have to do this if you do not want to.

If you do not want to do this it will not change any of the treatment you are getting from the Doctors.

Thank you for reading and listening to this.

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Hospital NHS Trust

UCL INSTITUTE OF CHILD HEALTH

DEVELOPMENTAL ENDOCRINOLOGY RESEARCH GROUP
CLINICAL & MOLECULAR GENETICS UNIT

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology
UCL Institute of Child Health
30 Guilford Street
London WC1N 1EH
Tel: 020 7905 2887
Email: j.achermann@ich.ucl.ac.uk



PARTICIPANT INFORMATION SHEET

**Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)**

Name of Principal Investigator: Dr John Achermann

Dear

You are being invited to take part in a research study into *Novel Mechanisms in Reproductive Biology*.

This project is based at UCL Institute of Child Health and is supported by The Wellcome Trust and/or European Union.

Before you decide whether to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully. Talk to others if you wish.

- Part 1 of this information sheet tells you the purpose of this study and what will happen if you take part
- Part 2 of this information sheet gives you more detailed information about the conduct of this study

Please feel free to ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part.

We thank you for taking the time to read this.

Sincerely

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Hospital NHS Trust

What is the aim of the study?

The aim of this study is to find new mechanisms involved in the development and function of the reproductive system. We hope to find new genes and proteins involved in reproductive biology, and to work out how variations in known genes and proteins have their effect.

Why have I been chosen?

We are approaching individuals attending our clinic with disorders of reproductive development.

Why is this study being done?

Within the past twenty years, we have started to understand some of the processes that control reproductive development and function in humans.

These processes are controlled by **proteins**, such as growth factors in the gonads (testis, ovary) or hormones in the blood. These proteins are encoded by **genes**, which are regions of each person's DNA blueprint or genetic code found on the **chromosomes**.

By analysing the proteins/hormones, genes and chromosomes of people with reproductive disorders in more detail we hope to find new factors involved in reproductive biology. We also want to improve our knowledge of those factors we already know are important, such as steroid hormone enzymes and receptors.

The results of our research will help us to understand reproductive development and function better. These findings could help us to improve the diagnosis and management of individuals and families with reproductive disorders.

Do I have to take part?

No. It is up to you whether or not to take part. If you do, you will be given this information sheet to keep and be asked to sign a consent form. You are still free to withdraw at any time and without giving a reason. A decision to withdraw at any time, or a decision not to take part, will not affect the standard of care you receive.

What will happen to me if I am interested in taking part?

If you agree to participate in the study, we will ask you to provide a small blood sample (two teaspoonfuls) or saliva sample (two teaspoonfuls, using a special kit) to extract DNA. This DNA will be stored and analysed for changes in specific genes related to reproductive biology. Some of these genes are currently known; others we hope to identify in the future. Wherever possible, we will take these blood samples at the same time that routine clinical tests are being done.

We might also ask you to provide additional blood samples (four teaspoonfuls, 20mls) to allow us to analyse hormones and proteins present in the blood stream, and to grow blood cells to perform more detailed analysis of chromosomes. We would be looking for small changes in chromosome structure that may not be picked up on routine clinical tests. Some of these detailed chromosome analysis studies can also be performed on DNA.

Usually only single samples are requested, although we may request an extra sample if we find something we would like to investigate in more detail.

We also request your consent to take urine samples for hormone profiles in some situations, and to have your approval to retain some surplus pieces of the gonad (testis, ovary) or skin at the time of an operation if this ever occurred to allow us to perform more detailed analysis of genes, proteins and hormones in these tissues.

With your consent, we plan to keep the samples for this current study and for other relevant studies in the future, if there is enough sample left and if we receive funding for such projects.

Your participation in this study will not require additional clinic visits, although if you would like us to tell you any of our findings this may require slightly longer outpatients appointments, or a special appointment arranged with myself or one of our clinical geneticists.

What are the alternatives for diagnosis or treatment?

The research tests proposed in this study are performed *in addition* to any standard clinical tests or investigations considered necessary as part of your routine clinical management. No routine tests or treatment would be withheld on the basis of whether you decide to participate in this study or not.

What are the potential risks and disadvantages of taking part?

No significant physical risk can be foreseen. We will try to take any blood samples at the same time that routine clinical tests are being performed, if possible. If a blood sample is being taken specifically for this study there will be some discomfort from the needle prick, but local anaesthetic cream will be used to numb the skin first. Issues related to genetic testing are discussed in more detail in Part 2.

What are the possible benefits of taking part?

There may be no immediate benefit to you from taking part in this study, but our research will help us to understand reproductive development and function better. This information may help others with conditions affecting the reproductive system or puberty.

Also, there is a small possibility that we might find out more information related to your specific condition. You will have the option whether any potential results are given to you. You can choose whether we give you an aggregate result (for example, 3 out of 20 participants had interesting findings) or whether we give you your actual results.

As this is a research study rather than an approved clinical “diagnostic” test, we would first check our findings on extra samples, and then repeat these studies in a clinical laboratory that undertakes genetic tests, if this is possible. The importance or implications of our research findings may not be known for some time, or clinical tests may not be readily available. It is very important to realize that our results are research findings and are not a clinical test. Further information related to genetic tests is provided in Part 2.

What happens when the research study stops?

We would like to keep any samples and relevant information related to this study for future research analysis beyond this current project. Undertaking further research work will be dependent on future funding.

What will happen if I don't want to carry on with the study?

Your participation in this study is voluntary. If you decide now or at a later stage that you do not wish to participate in this research project you are free to withdraw at any time, without giving any reason. Your medical treatment or legal rights will not be affected. Any stored samples will be destroyed if you wish.

What if there is a problem?

Any complaint about the way you have been dealt with during the study or any possible harm you might suffer will be addressed. The detailed information on this is given in Part 2.

Will my taking part in the study be kept confidential?

Yes. All information about your participation will be kept confidential. The details are included in Part 2.

Contact details:

For further information at any stage, you are welcome to contact: Dr John Achermann, Wellcome Trust Senior Research Fellow in Clinical Science, Endocrinology, UCL Institute of Child Health, 30 Guilford Street, London WC1N 1EH (0207 905 2887 or j.achermann@ich.ucl.ac.uk).

This completes Part 1 of the Information Sheet. If you are interested in participating, please continue to read the additional information in Part 2 before making any decision.

PART 2 – Detailed information

What if relevant new information becomes available?

Your participation in this research study will not interfere with you being offered any relevant new tests or management approaches that might become available in the future.

Will my taking part in this study be kept confidential?

All information that is collected about you during the course of the research will be kept strictly confidential and in compliance with the Data Protection Act 1998. Any paper records will be stored securely and any electronic records bearing “identifiers” (e.g., names) will be password protected. Samples in the Research Laboratory will be given a research number, which will be used during analysis rather than names.

Only the researchers and a representative of the Research Ethics Committee will have access to the data collected during the study. You will be asked to agree that appropriate sections of your medical notes may be looked at by responsible individuals from the Institute of Child Health where it is relevant to this research.

Notification of your General Practitioner/Family Doctor (GP) and specialists involved in your care

We will ask you for your permission to inform your GP and other specialists about your participation in this study. In some situations it will be important to discuss potentially significant research findings with other members of the clinical and research team, and other specialists involved in your care.

What will happen to any samples I give?

Any samples provided will be processed initially in the Clinical Genetics Laboratory of Great Ormond Street Hospital (DNA extraction), Regional Cytogenetics Laboratory (chromosomes), or in the Research Laboratory at the Institute of Child Health (salivary DNA, serum/plasma) before being transferred for storage and analysis by the research team involved in this study at the Institute of Child Health.

Everyone taking part in this study will be given a study number (“coded”) and samples kept in the research laboratory will be handled on a day-to-day basis using this number. Only the research team involved in this study will have access to these samples, and only the principal investigator and designated individuals will be able to identify whose sample it is.

We like to view any samples provided as “gifts” for the purposes of research. It is hoped that the samples will be kept for analysis by the research team on an ongoing basis, but it is quite possible that the samples will be used up during the course of this study. We also ask your permission to distribute coded samples and anonymous information to collaborators involved in related research within the UK, within the European Economic Area, and to countries outside of Europe. No names will be provided and you are free to choose whether you agree to this or not. With your consent, we may use some of the samples for commercial collaborations (i.e. partnerships between University and Industry) to develop better diagnostic and therapeutic tools. Commercial collaborators would not receive any identifying information, thus your personal information remains safe and confidential with the research team.

We do not anticipate that this work will lead to any outcome of commercial significance and you would not benefit financially if this research does lead to the development of a new treatment or medical test.

Will any genetic tests be done?

The aim of this study is to identify new genes involved in reproductive development and function, or variations in known genes involved in these systems.

We will be able to tell you some of the genes we plan to look at when you start the study. We will also ask you whether we can analyse additional factors that may be relevant in reproductive biology as they are identified. You may allow us to do this without contacting you further, or you may request we contact you to get your agreement each time.

Although this is a research study, we are willing to share any potential positive findings with you. We may ask a clinical geneticist to join us to explain any findings. If you would prefer not to know the results of any of our investigations then that is your choice. **It is very important to appreciate that our results are research**

findings rather than clinical tests. We recommend checking any positive findings in a separate sample, and – if you wish – trying to confirm our findings in a clinically approved laboratory. However, it is quite possible that such clinical tests are not readily available at the present time. It is also possible that the significance of our findings is unclear at present. Finally, it is not currently possible to analyse all genes and proteins in their entirety, so we might overlook subtle changes in our studies.

In a few instances, we are also trying to analyse many genes at once using a specially designed “gene chip”. This new technology might be useful in the future for obtaining more rapid results for patients. If we are interested in including your DNA in our study to test this new technology we will discuss it with you.

What will happen to the results of the research study?

Our aim is that the results of these studies will be presented to the scientific community and important advances will be published in peer-reviewed scientific and medical journals. This is the best way of having our work reviewed by experts in the field, and allowing other doctors and patients to benefit from our findings.

You will not be identified in any report or publication unless you have consented to the release of such information. Clinical data, biochemical data (e.g. hormone results) and radiological (e.g. X-rays, scans), histological or whole-organ images (e.g. pictures of the gonad) may be used. External photographs will not be used without specific consent.

What if there is a problem?

If you have a concern about any aspect of this study please discuss them in the first instance with the principal researcher (Dr John Achermann, 0207 905 2887) who will do their best to answer your questions. If the problems are not resolved, or you wish to comment in any other way, please contact the Research and Development Office, Institute of Child Health, 30 Guilford Street, London WC1N 1EH (Emma Pendleton, 020 7905 2179).

This project has been approved by an independent Research Ethics Committee who believe that it is of minimal risk to you. However, research can carry unforeseen risks and we want you to be informed of your rights in the unlikely event that any harm should occur as a result of taking part in this study.

This research is covered by a no-fault compensation scheme, which may apply in the event of any significant harm resulting to you from involvement in the study. Under this scheme it would not be necessary for you to prove fault. You also have the right to claim damages in a court of law. This would require that you prove fault on the Hospital/Institute and/or any manufacturer involved.

Who is organizing and funding the research?

This study is being organized by Dr John Achermann at the UCL Institute of Child Health and is being funded by The Wellcome Trust as part of a Senior Fellowship in Clinical Science as well as by the European Union.

Who has reviewed the study?

This study has been reviewed and approved by the Research Ethics Committees of UCL Institute of Child Health/Great Ormond Street Hospital NHS Trust (REC reference 05/Q0508/24; Study number 05BC13).

If you are interested in taking part you will be given a copy of this information sheet and any signed consent form to keep.

Please feel free to contact us if you have any further questions, or if your care moves to a different hospital or clinic.

We thank you for considering taking part and for taking the time to read this information sheet.

Contact:

Dr John Achermann
Wellcome Trust Senior Fellow in Clinical Science
Honorary Consultant in Paediatric Endocrinology
UCL Institute of Child Health, 30 Guilford Street
London WC1N 1EH
Tel: 020 7905 2887, Email: j.achermann@ich.ucl.ac.uk



Centre: Great Ormond Street Hospital NHS Trust/UCL Institute of Child Health

Study Number: 05BC13

Patient Identification Number for this study:

CONSENT FORM FOR PARENTS/GUARDIANS OF CHILDREN PARTICIPATING IN RESEARCH STUDIES

Title of Project: Novel mechanisms in reproductive biology (incorporating EuroDSD and UKGAIN)

Name of Principle Investigator: Dr John Achermann

Please initial box

Yes No

1. I confirm that I have read and understand the information sheet entitled "*Reproductive Biology Parent/Guardian Information 07August 2008 v3*" for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily by _____ Yes No
2. I understand that my child's participation is voluntary and that they are free to withdraw at any time without giving any reason, without his/her medical care or legal rights being affected. Yes No
3. I understand that relevant sections of any of my child's Medical Notes and data collected during the study may be looked at by employees from Regulatory Authorities or from Great Ormond Street Hospital/Institute of Child Health, where it is relevant to my child's taking part in this research. I give permission for these individuals to have access to my child's records. Yes No
4. I agree to my GP to be informed of my child's participation in the study. Yes No
5. I agree to my child taking part in the above study. Yes No

Name of Child

Date of Birth

Hospital number

Name of Parent/Guardian

Date

Signature

Name of Person taking consent
(if different from Investigator)

Date

Signature

Investigator

Date

Signature

One copy for parent/guardian; one copy for R&D section in the Medical Notes; original to be kept in the PI's site file

UCL Clinical & Molecular Genetics Unit

Institute of Child Health, 30 Guilford Street, London WC1N 1EH

Tel: +44 (0)20 7905 2887 Fax: +44 (0)20 7404 6191

e-mail: j.achermann@ich.ucl.ac.uk

Reproductive Biology Parent/Guardian Consent 07August2008 v3



Centre: Great Ormond Street Hospital NHS Trust/UCL Institute of Child Health

Study Number: 05BC13

Patient Identification Number for this study:

CONSENT FORM FOR PARENTS/GUARDIANS OF CHILDREN PARTICIPATING IN RESEARCH STUDIES

Title of Project: Novel mechanisms in reproductive biology/EuroDSD/UKGAIN

Name of Principle Investigator: Dr John Achermann

	Please initial box	
	Yes	No
6a. I agree to my child having a blood/saliva sample taken for DNA extraction.	<input type="checkbox"/>	<input type="checkbox"/>
6b. I agree to my child's DNA sample being analysed on a research basis for the following genes: _____.	<input type="checkbox"/>	<input type="checkbox"/>
6c. I agree to my child's DNA sample being stored for future analysis.	<input type="checkbox"/>	<input type="checkbox"/>
6d. I agree to my child's DNA sample being analysed on a research basis for other genes that may be related to reproductive biology:		
I) without further notification	<input type="checkbox"/>	<input type="checkbox"/>
OR		
II) I would like to be informed prior to additional analysis (if not in clinic, I would prefer telephone/letter/email [delete])	<input type="checkbox"/>	<input type="checkbox"/>
6e. I agree to my child's DNA being used to analyse multiple genes with the "Gene Chip"	<input type="checkbox"/>	<input type="checkbox"/>
6f. I agree to DNA/chromosome analysis for small rearrangements, deletions or duplications	<input type="checkbox"/>	<input type="checkbox"/>
7a. I would like to be informed of the results of any positive research findings.	<input type="checkbox"/>	<input type="checkbox"/>
7b. If yes (7a), I would like to receive any results in an aggregate format (for example, 3 out of 20 people assessed were found to have interesting results)	<input type="checkbox"/>	<input type="checkbox"/>
7c. If yes (7a), I would like to receive results specific to my child (please note, these are research findings and do <u>not</u> represent a clinically approved test)	<input type="checkbox"/>	<input type="checkbox"/>

Name of Child

Date of Birth

Hospital number

Name of Parent/Guardian

Date

Signature

Investigator

Date

Signature

One copy for parent/guardian; one copy for R&D section in the Medical Notes; original to be kept in the PI's site file

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Reproductive Biology Parent/Guardian Consent 07August2008 v3

UCL INSTITUTE OF CHILD HEALTH

DEVELOPMENTAL ENDOCRINOLOGY RESEARCH GROUP

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Dr John Achermann

Wellcome Trust Senior Research Fellow in Clinical Science

Honorary Consultant in Paediatric Endocrinology



Centre: Great Ormond Street Hospital NHS Trust/UCL Institute of Child Health

Study Number: 05BC13

Patient Identification Number for this study:

**CONSENT FORM FOR PARENTS/GUARDIANS OF CHILDREN
PARTICIPATING IN RESEARCH STUDIES**

Title of Project: Novel mechanisms in reproductive biology/Euro DSD/UKGAIN

Name of Principle Investigator: Dr John Achermann

Please initial box

Yes No

- 8a. I agree to my child having a blood sample(s) taken for serum/plasma extraction, stored and analysed for hormones and proteins that could be related to reproductive biology. Yes No
- 9a. I agree to my child having a urine sample(s) taken, stored and analysed for hormones and proteins that may be related to reproductive biology. Yes No
- 10a. I agree, where relevant, to some of my child's tissue samples (gonad/skin/fibroblasts cells/urine) being retained, stored and analysed if appropriate for genes, proteins and hormones that could be related to reproductive biology. Yes No
- 11a. I agree to my child's samples (for example, DNA, serum/plasma, tissue samples such as gonad/skin/fibroblast cells/urine) being kept and used in future ethically approved research projects. Yes No
- 12a. I agree to my child's clinical information and/or samples (for example, DNA, serum/plasma, tissue samples such as gonad/skin/fibroblast cells) being shared with researchers in other universities where appropriate. Data and/or samples sent to other international centres will be handled with the appropriate code of ethical conduct. Yes No
- 12b. I agree to my child's clinical information and/or samples (for example, DNA, serum/plasma, tissue samples such as adrenal/skin/fibroblast cells/urine) being shared with commercial collaborators where appropriate. Data and/or samples sent to other commercial entities will be handled with the appropriate code of ethical conduct. Yes No
- 13a. I agree to my child's clinical information and research results being used in scientific presentations and journal publications. No names or specific identifiers will be used. Additional consent will be obtained for the use of external photographs. Yes No

Name of Child

Date of Birth

Hospital number

Name of Parent/Guardian

Date

Signature

Investigator

Date

Signature

One copy for parent/guardian; one copy for R&D section in the Medical Notes; original to be kept in the PI's site file

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Reproductive Biology Parent/Guardian Consent 07August2008 v3



Centre: Great Ormond Street Hospital NHS Trust/UCL Institute of Child Health

Study Number: 05BC13

Patient Identification Number for this study:

**CONSENT FORM FOR PARTICIPANTS
IN RESEARCH STUDIES**

**Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)**

Name of Principle Investigator: Dr John Achermann

- | | Please initial box | |
|--|---------------------------|--------------------------|
| | Yes | No |
| 1. I confirm that I have read and understand the information sheet entitled " <i>Reproductive Biology Adult Information 07August 2008 v3</i> " for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily by _____. | <input type="checkbox"/> | <input type="checkbox"/> |
| 2. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care or legal rights being affected. | <input type="checkbox"/> | <input type="checkbox"/> |
| 3. I understand that relevant sections of any of my Medical Notes and data collected during the study may be looked at by employees from Regulatory Authorities or from Great Ormond Street Hospital/Institute of Child Health, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my records. | <input type="checkbox"/> | <input type="checkbox"/> |
| 4. I agree to my GP to be informed of my participation in the study. | <input type="checkbox"/> | <input type="checkbox"/> |
| 5. I agree to take part in the above study. | <input type="checkbox"/> | <input type="checkbox"/> |

Name

Date of Birth

Hospital number

Date

Signature

Name of Person taking consent
(if different from Investigator)

Date

Signature

Investigator

Date

Signature

One copy for participant; one copy for R&D section in the Medical Notes; original to be kept in the PI's site file

UCL INSTITUTE OF CHILD HEALTH

DEVELOPMENTAL ENDOCRINOLOGY RESEARCH GROUP

CLINICAL & MOLECULAR GENETICS UNIT

Dr John Achermann

Wellcome Trust Senior Research Fellow in Clinical Science

Honorary Consultant in Paediatric Endocrinology



Centre: Great Ormond Street Hospital NHS Trust/UCL Institute of Child Health

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IN RESEARCH STUDIES**

Title of Project: Novel mechanisms in reproductive biology/EuroDSD/UKGAIN

Name of Principle Investigator: Dr John Achermann

- | | Please initial box | |
|---|--------------------------|--------------------------|
| | Yes | No |
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| 6c. I agree to my DNA sample being stored for future analysis. | <input type="checkbox"/> | <input type="checkbox"/> |
| 6d. I agree to my DNA sample being analysed on a research basis for other genes that may be related to reproductive biology: | | |
| I) without further notification | <input type="checkbox"/> | <input type="checkbox"/> |
| OR | | |
| II) I would like to be informed prior to additional analysis (if not in clinic, I would prefer telephone/letter/email [delete]) | <input type="checkbox"/> | <input type="checkbox"/> |
| 6e. I agree to my DNA being used to analyse multiple genes with the "Gene Chip" | <input type="checkbox"/> | <input type="checkbox"/> |
| 6f. I agree to DNA/chromosome analysis for small rearrangements, deletions or duplications | <input type="checkbox"/> | <input type="checkbox"/> |
| 7a. I would like to be informed of the results of any positive research findings. | <input type="checkbox"/> | <input type="checkbox"/> |
| 7b. If yes (7a), I would like to receive any results in an aggregate format (for example, 3 out of 20 people assessed were found to have interesting results) | <input type="checkbox"/> | <input type="checkbox"/> |
| 7c. If yes (7a), I would like to receive results specific to myself (please note, these are research findings and do <u>not</u> represent a clinically approved test) | <input type="checkbox"/> | <input type="checkbox"/> |

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Date of Birth

Hospital number

Date

Signature

Investigator

Date

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Reproductive Biology Patient Consent 07August 2008 v3

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**CONSENT FORM FOR PARTICIPANTS
IN RESEARCH STUDIES**

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Name of Principle Investigator: Dr John Achermann

Please initial box

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- 10a. I agree, where relevant, to some of my tissue samples (gonad/skin/fibroblasts cells) being retained, stored and analysed if appropriate for genes, proteins and hormones that could be related to reproductive biology. Yes No
- 11a. I agree to my samples (for example, DNA, serum/plasma, tissue samples such as gonad/skin/fibroblast cells/urine) being kept and used in future ethically approved research projects. Yes No
- 12a. I agree to my clinical information and/or samples (for example, DNA, serum/plasma, tissue samples such as gonad/skin/fibroblast cells/urine) being shared with researchers in other universities where appropriate. Data and/or samples sent to other international centres will be handled with the appropriate code of ethical conduct. Yes No
- 12b. I agree to my clinical information and/or samples (for example, DNA, serum/plasma, tissue samples such as adrenal/skin/fibroblast cells/urine) being shared with commercial collaborators where appropriate. Data and/or samples sent to other commercial entities will be handled with the appropriate code of ethical conduct. Yes No
- 13a. I agree to my clinical information and research results being used in scientific presentations and journal publications. No names or specific identifiers will be used. Additional consent will be obtained for the use of external photographs. Yes No

Name

Date of Birth

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Date

Signature

Investigator

Date

Signature

One copy for participant; one copy for R&D section in the Medical Notes; original to be kept in the PI's site file

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Reproductive Biology Patient Consent 07August 2008 v3



ASSENT FORM FOR CHILDREN
(to be completed by the child and their parent/guardian)

Title of Project: Novel mechanisms in reproductive biology
(incorporating EuroDSD and UKGAIN)

Child (or if unable, parent on their behalf) /young person to circle all they agree with please:

- | | |
|--|--------|
| Have you read (or had read to you) about this project? | Yes/No |
| Has somebody else explained this project to you? | Yes/No |
| Do you understand what this project is about? | Yes/No |
| Have you asked all the questions you want? | Yes/No |
| Have you had your questions answered in a way you understand? | Yes/No |
| Do you understand it's OK to stop taking part at any time? | Yes/No |
| Are you happy to take part? | Yes/No |
| Are you happy for the doctors to keep and store samples (such as your blood, DNA, skin, or tissue) for use in this project and in future projects? | Yes/No |

If any answers are 'no' or you **don't** want to take part, **don't** sign your name!

If you do want to take part, please write your name and today's date

Your name _____

Date _____

Your parent or guardian must write their name here too if they are happy for you to do the project

Print Name _____

Sign _____

Date _____

The doctor who explained this project to you needs to sign too:

Print Name _____

Sign _____

Date _____

Thank you for your help.

(One copy for the Participant; one copy for the R&D section in the Medical Notes; original to be kept in the PI's site file)