FORORD

Denne rapporten beskriver arbeidet med prosjektet Europeisk Aniridikonferanse Oslo 2012, et forebyggingsprosjekt for å øke kunnskapen om Aniridi, som er en sjelden, medfødt genetisk øyelidelse.

Målet med prosjektet var å avholde en medisinsk konferanse om aniridi i Norge. Dette skjedde som planlagt i juni 2012.

Foreningen Aniridi Norge sto bak prosjektet. Dette er en liten diagnoseorganisasjon, som hadde 164 medlemmer på tidspunktet da søknaden ble sendt. Cirka en tredjedel av disse har aniridi.

Våre samarbeidspartnere har vært Norges Blindeforbund og Senter for sjeldne diagnoser ved Oslo Universitetssykehus (SSD). Vi har også samarbeidet tett med Huseby Kompetansesenter. Internasjonale samarbeidspartnere har vært Aniridia Europe, det europeiske nettverket av aniridiforeninger, og det amerikanske Aniridia Foundation International.

De som aller mest fortjener takk for hjelpen er de medisinske ekspertene som frontet konferansen faglig: Overlege Trine Prescott, Avdeling for medisinsk genetikk, Oslo universitetssykehus, overlege Charlotte von der Lippe, SSD, dr. med. Liv Drolsum ved Øyeavdelingen, Ullevål Universitetssykehus og øyelege dr. med. Ruth Riise.

Uten deres innsats, ingen konferanse!

SAMMENDRAG

Aniridi er en svært sjelden, medfødt genetisk øyelidelse. I Norge er det cirka 70 personer med diagnosen. Mennesker med aniridi ser ut som om de har store mørke øyne. Dette skyldes at iris i øyet mangler helt eller delvis. Dette er imidlertid bare den lettest synlige delen av lidelsen. I realiteten er dette en øyesykdom som rammer hele øyet og medfører synshemming og mange komplikasjoner som i alvorlige tilfeller kan gi blindhet ved feil eller mangelfull behandling.

Mange med aniridi har erfart at kunnskapen om tilstanden er mangelfull blant medisinsk personell. Foreningen Aniridi Norge så et konkret behov for å få satt spørsmål om aniridi på dagsordenen blant norske øyeleger. Foreningen bestemte seg derfor for å arrangere en internasjonal medisinsk konferanse om aniridi. Til konferansen inviterte vi de fremste øyelegene og genetikerne i verden til å komme og dele sin kompetanse med hverandre og knytte nettverk. Tanken var at konferansen skulle øke den samlede kunnskapen om aniridi, til glede både for norske aniridipasienter og pasienter i andre land.

Konferansen ble avholdt 8. – 10. juni 2012. En lang rekke medisinske eksperter kom til Oslo for å gi kolleger og brukere innblikk i sine spesialfelt og utveksle kompetanse.

KAPITTEL 1. BAKGRUNN OG MÅLSETTING

Foreningen Aniridi Norge er en liten diagnoseforening for pasienter med aniridi og deres pårørende. Foreningen ble stiftet i 2003. Fra starten var det klart at en sentral målsetting var å etablere nettverk internasjonalt og komme i kontakt med relevante forskningsmiljøer.
Aniridi er en svært sjelden, medfødt genetisk øyelidelse. I Norge er det cirka 70 personer med diagnosen. En regner med at det blir født omtrent 1 barn per år i Norge med aniridi, altså en forekomst på 1 per 40.000-50.000 fødsler.

Mennesker med aniridi ser ut som om de har store mørke øyne. Dette skyldes at iris i øyet mangler helt eller delvis. Manglende iris er imidlertid bare den lettest synlige delen av lidelsen. I realiteten er dette en øyesykdom som rammer hele øyet og medfører synshemming og mange komplikasjoner som i alvorlige tilfeller kan gi blindhet ved feil eller mangelfull behandling.
Nedsatt syn, lysømfintlighet og overhyppighet av grønn stær og hornhinnefordunkling er vanlig. Aniridi kan forekomme sammen med syndromtilstander som WAGR og Gillespie syndrom.

Behandling av både grå og grønn stær, samt hornhinnefordunkling, krever helt spesielle forholdsregler når pasienten har aniridi.
Mange medlemmer i foreningen hadde erfart at kunnskapen om tilstanden aniridi er mangelfull blant leger og andre medisinsk personell. Foreningen så et konkret behov for å få satt spørsmål om aniridi på dagsordenen blant norske øyeleger.

Ideen om en internasjonal faglig konferanse tok form tidlig. Allerede i 2005 deltok representanter fra Aniridi Norge på internasjonale aniridikonferanser i Chicago (USA) og Spania. Hensikten var å knytte kontakter og utveksle erfaringer.
Kontakten har senere blitt opprettholdt og foreningen har jevnlig sendt representanter til faglige samlinger både i Spania og USA, som er de to landene utover Norge (og etterhvert Italia) der aniridi har hatt godt etablerte organisasjoner med internasjonal kontaktflate.

Sommeren 2011 hadde Aniridi Norge tre representanter på den amerikanske aniridikonferansen. Denne gangen var målsettingen å verve foredragsholdere til Oslo-konferansen. Foreningens utsendte identifiserte og opprettet kontakt med aktuelle fagpersoner. Disse var fremstående øyeleger og vurdert blant de fremste ekspertene på aniridi i verden.

Fordi diagnosen aniridi er så sjelden og kunnskapen så beskjeden, var håpet å skaffe så interessante internasjonale foredragsholdere at norske øyeleger ville fatte interesse for å komme på konferansen.

At Norges Blindeforbund på et tidlig stadium ga forhåndstilsagn om økonomisk og organisatorisk støtte til konferansen, var en svært viktig forutsetning for at foreningen besluttet å gå videre med forberedelsene.

Konferansen ble planlagt med en kostnadsside på 1 147 400 kroner. Foreningen tok mål av seg til å finansiere deler av dette selv, og dessuten innhente noe ekstern støtte fra kilder som Norges Blindeforbund. Egne inntekter ble budsjettert til 250 000, slik at udekket behov var 897 400. Dette ble også søknadsbeløpet til ExtraStiftelsen.

De største utgiftspostene i budsjettet var reise og opphold for nasjonale og internasjonale foredragsholdere, dagpakker for konferansedeltagere og overnatting.

KAPITTEL 2. PROSJEKTGJENNOMFØRING OG METODE

Aniridi skyldes oftest en mutasjon på PAX6-genet som styrer utviklingen av menneskeøyet i fosterlivet.
Fordi økt kunnskap om genetikken kan bringe forståelsen av aniridi fremover og på sikt kanskje også bidra til å kurere tilstanden, ønsket fagkomiteen å gjøre konferansen til et møte mellom øyeleger (som er eksperter på øyets kammervinkler, netthinne, synsnerve og hornhinne) og genetikere (som forsker på hvilke gener som styrer hva og hva som faktisk skjer når genene går i arv eller muterer og styrer utviklingen av øyet i feil retning).

Oppdaterte øyeleger er en garanti mot feilbehandling , som i verste fall kan medføre blindhet.
Forskning kan fremme nye og mer effektive behandlingsformer. Dette ønsket vi å
stimulere til med konferansen.
Innenfor genetikken var målet at konferansen skulle bringe sammen fremstående genetikere som forsker på det aktive PAX6-genet slik at de kan informere om virkningene av genmutasjonen og utveksle kunnskap.

Til sammen mente vi at konferansen ville bli et svært godt forebyggingstiltak for norske pasienter med aniridi.

I tillegg ønsket vi at konferansen skulle gi et tilbud til brukere, synspedagoger og annet helsepersonell. Det kom tydelige signaler fra Norges Blindeforbund om at man hadde gode erfaringer fra andre sammenhenger med å blande fagpersonell og pasienter og pårørende for å bryte ned barrierene mellom forskningsmiljøene og pasientgruppen.

Også for SSD var dette aspektet viktig, ettersom et av SSDs viktigste oppdrag er å bringe informasjon ut til brukergruppene. Ut av dette kom ideen om å kombinere den faglige delen med en brukerdag.

Brukerdagen ble lagt inn som en tredje dag i konferanseprogrammet. Dette skulle være en åpen dag med foredrag tilpasset brukerne, en arena for erfaringsutveksling. I og med at diagnosen forekommer så sjelden er det mange med aniridi som ikke har andre i sin nærhet med samme diagnose.

Dette skulle vise seg å bli en suksessfaktor. SSD påtok seg en sentral rolle i den praktiske gjennomføringen av brukerdagen.

Konferansen skulle avholdes over tre dager på forsommeren 2012. Det faglige programmet gikk over to dager med presentasjoner fra 15-20 spesialister. Tredje dag ble en åpen dag med foredrag tilpasset brukere, synspedagoger og annet helsepersonell.
På forhånd trodde vi at en faglig sterk konferanse vil kunne tiltrekke cirka 100 deltagere i tillegg til foredragsholdere.

Den praktiske planleggingen av konferansen startet umiddelbart etter tildeling av Extramidler mot slutten av 2010. Oppstartsmøtet for programkomiteen og arrangementskomiteen ble avholdt tirsdag 18. januar 2011. Den mest arbeidsintensive perioden løp fra mai 2011 helt frem til konferansen ble avholdt i Oslo 8. – 10. juni 2012.

I planleggingen av konferansen etablerte vi flere komiteer som ivaretok ulike oppgaver.
Norges Blindeforbund var hele veien med som støttespiller og rådgiver i en uformell styringsgruppe.

For å drive planleggingen fremover ble det nedsatt en arbeidsgruppe som besto av

- Asbjørn Akerlie, prosjektleder og styremedlem Aniridi Norge
- Hilde R. Hansen, leder Aniridi Norge
- Bjørn Eckblad, pårørende
- Arvid Meløy, styremedlem Aniridi Norge, har selv aniridi
- Sølvi Ørstenvik, styremedlem Aniridi Norge, pårørende
- Åse Hege Bratli, styremedlem i Aniridi Norge, pårørende

Fagkomiteen som påtok seg det medisinske ansvaret besto av:

- Overlege Trine Prescott, Avdeling for medisinsk genetikk, Oslo universitetssykehus.
- Dr. med. Liv Drolsum ved Øyeavdelingen, Ullevål Universitetssykehus
- Øyelege dr. med. Ruth Riise
- Trine Prescott ble senere erstattet av overlege Charlotte von der Lippe ved SSD.

Også representanter for Aniridi Norge møtte i fagkomiteen.

Det ble også nedsatt en egen komité med ansvar for brukerprogrammet på dag tre. Den besto av:

- Asbjørn Akerlie (leder, pårørende)
- Silje Vangsnes (har aniridi)
- Sissel Brøndmo (synspedagog, Huseby kompetansesenter)
- Line Merete Mediå, (sykepleier, SSD)
- Elisabeth Daae (psykolog, SSD)
- Charlotte von der Lippe (overlege, SSD).

Denne komiteen hadde ansvar for å tilrettelegge dag 3, der brukerne fikk et sammendrag av de viktigste medisinske presentasjonene fra de to foregående dagene. Det var også anledning til å stille spørsmål til forskerne i en egen sesjon.

Til å ivareta det arrangementstekniske leide vi inn en profesjonell konferansearrangør, Meeting Management. Deres primære oppgave var administrasjon av booking av hotell og registrering av konferansedeltagere.

Det ble også bestemt at det skulle avholdes en synsklinikk som en del av det faglige programmet under konferansen. Her ble det etablert et samarbeid med Huseby kompetansesenter. Alle brukere som var interessert, fikk anledning til å bli undersøkt på Huseby kompetansesenter av en gruppe bestående av de fremste, internasjonale ekspertene på aniridi, glaukoma, hornhinne.

Pr- og markedsføring
Vi drev utstrakt markedsføring av konferansen både gjennom annonser i Tidsskrift for den norske legeforening og på relevante nettsteder. Informasjon om konferansen ble også distribuert i sosiale medier, som for eksempel på Facebook-sidene til relevante legeforeninger. Konferansen ble også kunngjort internasjonalt på relevante faglige nettsteder.

En egen engelskspråklig nettside ga nødvendig informasjon om konferanseprogram, påmelding og praktiske opplysninger. http://www.aniridiaconference.org/

KAPITTEL 3. RESULTATER OG RESULTATVURDERING

Målsettingen med konferansen var å bringe sammen de fremste ekspertene på behandling av aniridi slik at de kunne utveksle informasjon og knytte kontakter.
Vi ønsket å belyse aniridi bredt, og inviterte foredragsholdere innen både øyemedisin og genetikk, med fokus på forebyggende tiltak og konkret behandling av tilleggskomplikasjoner. Sosiale og psykologiske utfordringer knyttet til diagnosen ble også berørt.

Til sammen deltok 50 medisinske eksperter under konferansen. Disse fordelte seg på:

21 inviterte foredragsholdere fra Norge, Sverige, Storbritannia og USA
24 påmeldte fagpersoner fra Norge, Canada, Belgia, Sverige og Italia
Fem medlemmer i fagkomiteen og arrangørkomiteen, som alle var norske

I tillegg meldte til sammen 58 pasienter, pårørende og ledsagere og tolker seg til hele eller deler av konferansen.

Vi ønsket å arrangere en europeisk aniridikonferanse for å legge til rette for at brukere med aniridi får best mulig medisinsk forebygging, behandling og oppfølging. Fordi det er få tilfeller i hvert land, så vi stor nytte av å arrangere en større europeisk, medisinsk konferanse.
Hensikten var å gjøre øyeleger i stand til å yte bedre hjelp gjennom kompetanseutveksling på tvers av grensene. Dette vil hindre unødig synstap for flest mulig barn, voksne og eldre med diagnosen.
Et sentralt siktemål var å stimulere til mer forskning, ikke minst blant norske øyeleger, fordi forskning kan fremme ny og mer effektiv behandling.

Å samle så mange eksperter rundt temaet aniridi var det viktigste vi oppnådde. Under konferansen kunne vi konstatere at forskerne utvekslet funn og innsikter, og dessuten fikk anledning til å treffe hverandre i en uformell, sosial setting. Hva som konkret kommer ut av dette på lengre sikt, er naturligvis vanskelig å fastslå i dag. Vårt klare inntrykk er imidlertid at konferansen bidro til å styrke interessen for vår sjeldne diagnose.

Flere yngre forskere presenterte sine arbeider, og ble innlemmet i et fellesskap av både yngre og mer erfarne vitenskapsmenn- og kvinner. Det ble luftet muligheter for finansiering og samarbeid om ytterligere forskning, med aniridi som samlende punkt.

En oversikt over alle de faglige presentasjonene som ble holdt, ble samlet i et eget hefte som alle deltagere mottok. Heftet ligger vedlagt til denne rapporten.

Videre ble ideen om å arrangere en ny, europeisk aniridikonferanse i Venezia i Italia i 2013 unnfanget. Lykkes dette, er kontinuiteten sikret og verdien av nettverkene som ble etablert i Oslo vil bli enda større.

For brukerne som deltok, ga foredragene økt innsikt i de mange komplekse og ubesvarte spørsmålene om hvorfor og hvordan aniridi oppstår og hva tilstanden bringer med seg av utfordringer gjennom livet. Ikke minst fikk de med seg noen spennende vyer om hvilke mulige nyvinninger innenfor behandling og terapi forskerne arbeider med.

For prosjektgruppen og søkerorganisasjonen har arbeidet med prosjektet vært et voldsomt løft. Vi har vist at vi er i stand til å hente ut uante krefter og få folk til å samarbeide for felles interesser.

Bevilgningen fra Extrastiftelsen var totalt på 897 000 kroner. I konferanseåret 2012 mottok prosjektet 837 000 kroner i støtte. Av dette brukte prosjektet 790 729 kroner, slik at 193 447 kroner står ubrukt og vil bli tilbakebetalt.

Vi mener prosjektet har betydelig overføringsverdi til andre sjeldne diagnoseforeninger, som ser behov for å samle kompetanse på tvers av landegrenser. Prosjektet viser at det er mulig å arrangere en internasjonal konferanse der man samler den beste ekspertisen som er tilgjengelig, og bringer den til Norge for å møtes og utveksle kunnskap.

Vi tenker at den viktigste jobben med å distribuere kunnskap og etablere kontakter skjedde under konferansen. Det ble både arrangert uformelle samlinger på kveldstid, en middag for alle de inviterte foredragsholderne og en bankett for alle deltagere. Her kunne både brukere og eksperter møtes og bli bedre kjent utenfor foredragssalen.

KAPITTEL 4. OPPSUMMERING, KONKLUSJON OG VIDERE PLANER

Aniridi Norge vil bestrebe seg på å opprettholde og videreutvikle kontakten med forskermiljøet, både nasjonalt og internasjonalt. Konkret vil dette skje gjennom deltagelse på konferanser i Norge og i utlandet. Vi vil også bistå den italienske aniridiforeningen i dens arbeid med en mulig konferanse i Italia i 2014. Håpet er at en kan etablere en fast europeisk konferanse som avholdes i et nytt land hvert annet år. Slik vil aniridimiljøet i Europa opprettholde og videreutvikle tilknytningen til forskermiljøene. Gjennom en slik tett tilknytning er forutsetningen til stede for å stimulere til ny forskning og kompetanseutveksling som vil komme alle med aniridi til gode.

Fremtidsutsiktene blir bedre for alle med aniridi hvis vi lykkes i dette.

Prosjektorganisasjonen avslutter sitt arbeid med denne sluttrapporten. Men erfaringene og nettverket blir værende i det norske aniridimiljøet og i Aniridi Norge.

VEDLEGG:

Extra - Regnskap 2012
Revisorberetning konferanse 2012
Booklet print - endelig versjon.pdf
20120323 NY Medisinsk pressemelding.pdf
20120323 Medical press relase English
Lauderdale en slide (eksempel på faglig presentasjon)
Veroncia von Heyningen Slide 2 (eksempel på faglig presentasjon)
20120608 Pressemelding Aniridikonferansen 2012
2012-03-23 Skjermbilde Overlegeforeningen (eksempel på markedsføring i sosiale medier)
2012-03-23 Skjermbilde Legeforeningens kurskatalog I (eksempel på markedsføring)
2012-03-15 Skjermbilde Helsedirektoratet (eksempel på markedsføring)
http://www.aniridiaconference.org/

ExtraStiftelsen
Helse og Rehabilitering

Regnskapsrapportering
ExtraStiftelsen

Meløy, Arvid

Kvittering

Opplysningene er sendt, ditt referansenummer er: A2XNKQ

Det er gjennomført en maskinell kontroll under utfylling, men vi tar forbehold om at det kan bli oppdaget feil under saksbehandlingen og at annen dokumentasjon kan være nødvendig.
Vennligst oppgi fødselsnummer/organisasjonsnummer og referansenummer ved eventuelle henvendelser.

Takk for samarbeidet.

Les mer om kvittering.

Avslutt

Sammendrag

Prosjektinformasjon

Spørsmål	Ditt svar
Virksomhetsområde	Forebygging

Prosjektinformasjon

Spørsmål	Ditt svar
Prosjektnummer	2010/1/0028
Prosjektnavn	Europeisk Aniridikonferanse Oslo 2012

Prosjektleder

Spørsmål	Ditt svar
Fornavn	Asbjørn
Etternavn	Akerlie

Prosjektets startdato

Spørsmål	Ditt svar
Registrert startdato på dette prosjektet er satt til	01.01.2011
Ble prosjektet startet denne datoen?	Ja

Prosjektets sluttdato

Spørsmål	Ditt svar
Registrert sluttdato på dette prosjektet er satt til	15.06.2012
Ønsker du å endre denne datoen?	Nei

Regnskap

VIKTIG OM REGNSKAPSRAPPORTERING:Når skjemaet er lagret eller sendt generes det et oppdatert regnskapsskjema i pdf-versjon. Dette åpnes via pdf-ikonet i Innboksen og i mappa Regnskapsskjema, skrives ut, undertegnes av prosjektleder, og sendes til revisor. Les mer informasjon og krav til regnskapsrapporteringen her.

Skriv inn tallene i formatet: xxxx eller xxxx,xx

Spørsmål	Ditt svar	
Regnskap for år	2012	
Inntekter		
Bevilget til prosjektet dette regnskapsåret	837000	
Ubrukte midler fra forrige regnskapsår	58058	
Offentlige midler		Spesifisering
Andre inntekter	89118	Egenandeler deltagere
SUM inntekter	984176	
Egne midler		
Totalsum	984176	
Utgifter	Beløp i kr.	
Lønn, sosiale utgifter	56732	
Innkjøpte tjenester/honorarer	109159	
Materiell/ utstyr		Spesifisering
Andre prosjektutgifter	595883	Reise og opphold
	28955	Tlf./Imternett, møter
SUM utgifter	790729	
Gjenstår evt. ubrukt per 31.12	193447	
Totalsum	984176	

Revisorkollegiet
- mer personlig

REVISJONSBERETNING FOR PROSJEKTREGNSKAP FINANSIERT AV EXTRASTIFTELSEN 2012

PROSJEKTNUMMER: 2010/1/0028

PROSJEKTNAVN: Europeisk Aniridikonferanse Oslo 2012

SØKERORGANISASJON: Aniridi Norge

REGNSKAPSPERIODE: **1.1.2012-31.12.2012**

Uttalelse om prosjektregnskapet

Vi har revidert prosjektregnskapet for ovennevnte prosjekt som består av resultatregnskap som viser totale kostnader på kr **790 729** og opplysning om ubrukte midler som utgjør kr **193 447**. Prosjektregnskapet er utarbeidet av ledelsen og prosjektleder/forsker
basert på regnskapsprinsippene i prosjektregnskapet som beskriver grunnlaget for utgiftsoppstillingen, og regler for regnskapsrapportering publisert på www.extrastiftelsen.no/til-mottakere/regnskap-og-rapportering og i brev av desember 2012.

Ledelsens ansvar for prosjektregnskapet

Ledelsen er ansvarlig for utarbeidelsen av prosjektregnskapet, og for slik intern kontroll som ledelsen finner nødvendig for å muliggjøre utarbeidelsen av et prosjektregnskap som ikke inneholder vesentlig feilinformasjon, verken som følge av misligheter eller feil.

Revisors oppgaver og plikter

Vår oppgave er å gi uttrykk for en mening om prosjektregnskapet på bakgrunn av vår revisjon. Vi har gjennomført revisjonen i samsvar med lov, forskrift og god revisjonsskikk i Norge, herunder International Standards on Auditing og veiledning for revisorer publisert på www.extrastiftelsen.no/til-mottakere/regnskap-og-rapportering. Disse reglene beskriver også følgende dokumenter som grunnlag for revisors kontroll:

- Prosjektsøknad med opprinnelig budsjett
- ExtraStiftelsens forutsetning/kommentar til bevilgningen
- Godkjente endringer fra ExtraStiftelsen som påvirker opprinnelig budsjett

Revisjonsstandardene krever at vi etterlever etiske krav og planlegger og gjennomfører revisjonen for å oppnå betryggende sikkerhet for at prosjektregnskapet ikke inneholder vesentlig feilinformasjon.

En revisjon innebærer utførelse av handlinger for å innhente revisjonsbevis for beløpene og opplysningene i prosjektregnskapet. De valgte handlingene avhenger av revisors skjønn, herunder vurderingen av risikoene for at prosjektregnskapet inneholder vesentlig feilinformasjon, enten det skyldes misligheter eller feil. Ved en slik risikovurdering tar revisor hensyn til den interne kontrollen som er relevant for virksomhetens utarbeidelse av et prosjektregnskap som gir uttrykk for prosjektets resultat i samsvar med regnskapsprinsippene beskrevet i prosjektregnskapet og regler for regnskapsrapportering publisert på www.extrastiftelsen.no/til-mottakere/regnskap-og-rapportering. Formålet er å utforme revisjonshandlinger som er hensiktsmessige ut fra omstendighetene, men ikke for å gi uttrykk for en mening om effektiviteten av enhetens interne kontroll. En revisjon omfatter også en vurdering av om de anvendte regnskapsprinsippene er hensiktsmessige, samt en vurdering av den samlede presentasjonen av prosjektregnskapet.

Revisorkollegiet AS
Statautoriserte revisorer
Medlemmer av Den norske Revisorforening

Revisornr./org.nr.:
987 593 636 MVA

post@revisorkollegiet.no
www.revisorkollegiet.no

PARTNERE:
Svein Andersen
Øivind Andersson
Petter B. Andreassen
John Arild Delsbekk
Terje Gulliksen
Roger Larsen
Truls Kristian Syvertsen

DRAMMEN:
Buskerudveien 72
Postboks 818
3007 Drammen
Tlf.: 32 27 63 00
Fax: 32 27 63 01

KONGSBERG:
Nymoens Torg 11
3611 Kongsberg
Tlf.: 32 72 28 80
Fax: 32 72 28 81

ÅMOT:
Industriveien 38
Postboks 79
3341 Åmot
Tlf.: 32 77 87 70
Fax: 32 77 87 71

Etter vår oppfatning er innhentet revisjonsbevis tilstrekkelig og hensiktsmessig som grunnlag for vår konklusjon.

Konklusjon

Etter vår mening gir dette prosjektregnskap for 2012, i det alt vesentlige uttrykk for prosjektets resultat og stilling i samsvar med grunnlaget for regnskapsavleggelse beskrevet i prosjektregnskapet og regler for regnskapsrapportering publisert på www.extrastiftelsen.no/til-mottakere/regnskap-og-rapportering.

Uttalelse om overholdelse av avtalevilkår

Konklusjon

Basert på vår revisjon av prosjektregnskapet som beskrevet ovenfor, mener vi at søkerorganisasjonen har overholdt avtalevilkårene om bruk av prosjektmidlene i overensstemmelse med opprinnelig budsjett justert for endringer godkjent av ExtraStiftelsen, herunder at utstyr/materiell er anskaffet i overensstemmelse med budsjettvilkårene for bevilgningen og at kostnadene enkeltvis eller totalt ikke overstiger opprinnelig budsjett justert for endringer godkjent av ExtraStiftelsen.

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1st European Conference on Aniridia

Programme / Abstracts

Oslo, June 8th – 10th, 2012

Oslo University Hospital

ExtraStiftelsen
Helse og Rehabilitering

Aniridia Europe

Norges Blindeforbund
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Aniridia Foundation International

Aniridia Europe

Programme

Friday June 8th, 12.30 – 17.30

Time	Speaker/Activity	Subject
10.00 – 12.15	Registration	Coffee, tea, light refreshments, Load presentations
11.30 – 12.15	**LUNCH**	
12.30 – 12.40	C. von der Lippe	Welcome
12.40 – 13.10	A. Moore	The spectrum of eye disease associated with PAX6 mutations
13.10 – 13.40	I. Gottlob	Foveal morphology and eye movements in patients with PAX6 mutations
13.40 – 14.10	C. Riemann	Aniridia: A Vitreoretinal Perspective
14.10 – 14.30	**BREAK**	
14.30 – 14.50	J. Nerby	Living with aniridia
14.50 – 15.10	U. Eden	Epidemiology and 8 years follow-up study of Scandinavian aniridia patients
15.10 – 15.40	M. Yogarajah	PAX6 and the brain
15.40 – 16.10	D-E. Bamiou	Auditory processing and PAX6 (videopresentation)
16.10 – 16.30	All speakers	Questions / Discussion
19.30	Reception	

Saturday June 9th, 09.00 - 17.30

Time	Speaker/Activity	Subject
08.00 – 09.00		Load presentations, coffee and tea
09.00 – 09.05	C. von der Lippe	Welcome
09.05 – 09.35	V. van Heyningen	Mutational spectrum for aniridia, genotype-phenotype correlations, and the lessons for understanding gene function
09.35 – 09.55	J. Lauderdale	PAX6 and limbal stem cells
09.55 – 10.15	D. FitzPatrick	Aniridia with no detectable mutation at the PAX6 locus
10.15 – 10.45	M. Carstens Moe	Ex vivo expanded autologous limbal epithelial cells on amniotic membrane using a culture medium with human serum as single supplement
10.45 – 11.00	**BREAK**	
11.00 – 11.20	E. Simpson	Aniridia Gene Therapy: Pre-Clinical Studies
11.20 – 11.40	L. Drolsum	Cataract and corneal surgery in aniridia
11.40 – 12.40	4 speakers	Submitted abstracts
12.40 – 13.45	LUNCH	
13.45 – 14.05	P. Fagerholm	The surface of the eye in aniridia
14.05 – 14.25	J. Freeman	Aniridic Keratopathy: causes and treatments
14.25 – 14.50	P. Netland	Glaucoma in aniridia
14.50 – 15.05	**BREAK**	
15.05 – 15.35	K. Colby	The Boston Keratoprosthesis for aniridia: avoiding complications and optimizing outcomes
15.35 – 16.35	4 speakers	Submitted abstracts
16.35 – 17.15	All speakers	Questions / discussion
20.00	**Banquet**	

Sunday June 10th, 09.00 - 17.30

Time	Speaker/Activity	Subject
08.00 – 09.00	Registration	Load presentations, coffee and tea
09.00 – 09.10	L. Mediå	Welcome
09.10 – 11.00	Panel	Speakers from Friday and Saturday
11.00 – 11.15	BREAK	
11.15 – 11.35	K. Brandsborg	Difficult issues: how to talk to children (in Norwegian)
11.35 – 11.55	H. T. Ryen	Work – how to prepare (in Norwegian)
11.55 – 12.10	BREAK	
12.10 – 12.30	M. Almbakk	The challenging teenage years (in Norwegian)
12.30 – 12.50	U. Eden	The eye and aging in aniridia (in Swedish)
12.50 – 13.00	Course ends	
13.00	LUNCH	

Invited speakers Friday / Saturday

Professor A T Moore MA, FRCS, FRCOphth, FMedSci.

Anthony T Moore
Professor of Ophthalmology
UCL Institute of Ophthalmology and
Honorary Consultant Ophthalmologist
Moorfields Eye Hospital and Hospital for Children
Great Ormond Street London.

Tony Moore graduated from Oxford University Medical School in 1975 and after junior posts in Ophthalmology at St Bartholomew's Hospital in London and the Radcliffe Infirmary in Oxford moved to the residency training programme at Moorfields Eye Hospital in London. He then undertook a two-year fellowship in Paediatric Ophthalmology at the Hospital for Sick Children in London and the Hospital for Sick Children in Toronto. After his return to the U.K. he was appointed Lecturer in Clinical Ophthalmology, a post held jointly at the UCL Institute of Ophthalmology and the Institute of Child Health in London. In 1986 he moved to Addenbrooke's Hospital in Cambridge as a consultant with a special interest in paediatric ophthalmology and inherited eye disease. In 2001 he moved back to London when he was appointed to the Duke Elder Chair of Ophthalmology at the UCL Institute of Ophthalmology, London. He has honorary consultant appointments at Moorfields Eye Hospital and the Hospital for Children, Great Ormond Street, London where he has his clinical practice. His main research interests are in inherited eye disease particularly those affecting the retina

The spectrum of eye disease associated with *PAX6* mutations

Abstract
Mutations in the *PAX6* gene were first identified in patients with classical aniridia, both the familial and sporadic forms; the mutations were predicted to result in a non functional protein. Chromosomal deletions of 11p that encompass the PAX6 gene also result in classical aniridia. The mechanism of failure of development of the eye was thus thought to be due to haploinsufficiency. Later a wider spectrum of mutations in *PAX6,* including missense mutations and mutations, which removed the normal stop codon, resulting in a larger protein than normal were identified. These mutations are associated with a larger range of phenotypes, including isolated foveal hypoplasia, autosomal dominant keratitis, microphthalmos, optic disc anomalies and exudative retinopathy. This talk will discuss the range of developmental ocular phenotypes seen in association with mutations in the *PAX6* gene in man and will also discuss the later ophthalmic complications that can develop such as glaucoma, cataract and corneal stem cell failure. The talk will set the scene for later talks of specific aspects of the PAX6 phenotype.

Professor Irene Gottlob

Professor Gottlob is Austrian citizen and married with two children. She graduated from the Medical School of the University of Vienna, Austria, where she also completed her training in Ophthalmology. She spent three years of research in the physiology of the visual system at the University of Vienna and then at the Max-Planck Institute for Physiological and Clinical Research in Bad Nauheim, Germany. During this time she became greatly attracted to the study of the connection between the eyes and the brain. She then undertook clinical and research fellowships in Paediatric Ophthalmology, Neuro-Ophthalmology and Oculoplastic Ophthalmology at Wills Eye Hospital in Philadelphia, USA. She obtained the Habilitation (Univ. Doz title) at the University of Vienna in 1990. Before she was appointed in 1999 as Professor and Chair in Ophthalmology at the University of Leicester she was Head of Department of Strabismus and Neuro-ophthalmology at St. Gall, Switzerland.

Her main areas of research are amblyopia, the regulation of eye movements and their disorders, in particular nystagmus, foveal development and the genetics of neuro-ophthalmic diseases. Professor Gottlob is the author of more than 130 peer reviewed research papers and book chapters. She is section editor of the British Journal of Ophthalmology and on the editorial board of other scientific journals. Professor Gottlob is an enthusiastic teacher of students and trainees. She greatly enjoys teaching and has been named academic role model for students and trainees by the BMA.

Foveal morphology and eye movements in people with *PAX6* mutations

Abstract

Purpose: To investigate foveal hypoplasia and nystagmus in patients with PAX6 mutations and to compare them to other patients with nystagmus/and or foveal hypoplasia and to investigate whether characteristics on OCT correlate with visual acuity.

Methods: Patients with PAX6 mutations were compared using eye movement recordings and ultrahigh resolution spectral optical coherence tomography to patients with other forms of nystagmus such as idiopathic nystagmus, and nystagmus associated to albinism and achromatopsia.

Results: In subjects with PAX6 mutations nystagmus form and direction was highly variable even within families. The nystagmus had large amplitudes, mainly pendular waveforms and 80% of patients had predominantly vertical nystagmus. Foveal hypoplasia was similar as in albinism. Grading of retinal structures on OCT was correlated with visual acuity.

Discussion: Interestingly nystagmus characteristics are different in patients with PAX6 mutations from other patients with foveal hypoplasia such as albinism. OCT scans can help to predict visual acuity. It would be interesting to see whether OCT scans in young children can predict later visual acuity.

Christopher D. Riemann, M.D.

Christopher D. Riemann, M.D. obtained his undergraduate degree in Biomedical Engineering from John Hopkins University Whiting School of Engineering in February 1989. After a six-month period as a Chesapeake Research Foundation Fellow at the National Institute on Aging, he attended University of Maryland School of Medicine in Baltimore. While in medical school, Dr. Riemann performed independent research as a National Institutes of Health Medical Student Research Fellow (1990) as well as an American Heart Association Research Fellow (1991-92). He received a Maryland Senatorial Scholarship in 1991 and served as the Vice President of the University of Maryland Chapter of the American Medical Student Association. Dr. Riemann obtained his medical doctoral degree in the Spring of 1994.

Subsequently Christopher D. Riemann undertook a transitional internship at York Hospital in York, Pennsylvania, and completed Residency training in Ophthalmology at the Cleveland Clinic Foundation in Cleveland, Ohio. He then continued his training at the Cleveland Clinic Foundation as a vitreoretinal surgery Fellow where he developed expertise in both the medical and surgical management of retinal disorders.

Dr. Riemann is a member of the Vitreous Society, American Academy of Ophthalmology, Ohio State Medical Association, Cincinnati Academy of Medicine, Cincinnati Ophthalmology Society, the Association for Research in Vision and Ophthalmology, and the American Medical Association. His original research in the fields of Ophthalmology, Cardiology, and Endocrinology has been published in international peer reviewed scientific journals and has been presented at national scientific meetings.

In collaboration with the other Retinal Surgeons at Cincinnati Eye Institute, Dr. Riemann is a principal investigator or co-investigator for many phase two and phase three FDA trials.

Dr. Riemann specializes in medical and surgical vitreoretinal diseases including diabetic retinopathy, macular degeneration, retinal detachment, retinopathy of prematurity, vascular diseases of the retina, uveitis, histoplasmosis, complications of anterior segment surgery, endoscopic posterior segment surgery, and ocular trauma.

Aniridia - A vitroretinal Prespective

Abstract
Aniridia is a difficult and challenging disease to manage for multiple reasons. Posterior segment involvement in the form of foveal hypoplasia nearly always contributes to visual loss

in these patients and presents a barrier to early detection of serious ocular disease. Severe posterior segment involvement with aniridic fibrosis syndrome and secondary retinal detachment, ciliary body shut down and hypotony may lead to catastrophic visual loss, phthisis and complete loss of the globe. We will explore a rational way to think about aniridic patients with posterior segment disease, useful diagnostic ancillary testing and specialized surgical techniques which can offer even the most difficult patients a chance at preserving vision.

Jill Ann Nerby

Jill Ann Nerby
Congenital Eye Disorder Manager, Department of Ophthalmology
Director and Founder, Aniridia Foundation International
University of Virginia School of Medicine
Charlottesville, Virginia USA

Jill Ann Nerby founded this unique 501(c)3 non-profit organization which was started in 2001. In 2011, Aniridia Foundation International was invited to move their headquarters to the University of Virginia - Department of Ophthalmology and expand AFI's research program, medical conferences and patient care services.

Jill was born with sporadic Aniridia Syndrome and glaucoma which has brought her many challenges in life. According to many people and physicians, it is those experiences that have made her the passionate communicator, educator and inspirational person she is today. Jill's knowledge of this eye and medical syndrome, and actually living it every day, is the very reason she can relate to both physicians, researchers, as well as, patients and their families. This, combined with her passion to help others, and make a difference in the low vision population, caused her to create this successful non-profit organization that has been quickly embraced by the medical and research community.

Since 2000, Jill has been undergoing surgeries for glaucoma valve implants, stem cell allografts, corneal transplants, and other eye surgeries all while running this large organization and being a single mom to her son Michael, who also has aniridia and glaucoma due to familial inheritance.

Jill graduated college with a Bachelor of Science degree (Psychology and Biology) and finished her pre-med coursework while working in the field of occupational therapy in anticipation of going to medical school. Unfortunately, the perils of the corneal scarring and with no correctional treatment at the time, her dream of going into the medical field had to be augmented. Today, Jill is the Congenital Eye Disorder Manager in the Department of Ophthalmology at University of Virginia School of Medicine and Executive Director of Aniridia Foundation International. Jill is an international invited guest speaker and also exhibits at many medical or research meetings and Lions conventions. She has co-authored "Aniridia and WAGR Syndrome: A Guide for Patients and their Families" published by Oxford University Press. It is sold worldwide and was reviewed by a medical journal as being a great tool for medical, optometry and ophthalmology students too.

Jill with the help of the AFI medical and scientific advisors also created the International Aniridia Medical Registry and gene bank with the hope of advancing research and patient care for ***everyone*** who experiences the same conditions as those children and adults born with Aniridia Syndrome. While being born with underdeveloped eye structures and having multiple ocular and medical issues in one person is unusual, ***the conditions which make up Aniridia Syndrome are COMMON in the general population*** such as glaucoma, corneal

disease, cataracts and diabetes. With the assistance of Aniridia Foundation International's Medical and Scientific boards and participating universities, AFI collects extensive medical data, DNA sequencing results, clinical photographs, and medical tests to advance research for **both** the general public and those with Aniridia who live with these same common conditions.

Living with Aniridia

Abstract
Jill Ann Nerby was born with sporadic aniridia and glaucoma. She will be sharing her very interesting journey in life, her accomplishments, conditions she developed, and the surgeries, transplants and experimental procedures in efforts to maintain her vision. Yet all this has not stopped her from living...she did it all "living with aniridia".

Ulla Edén, MD, PhD

Dept. of Ophthalmology, Inst f Clinical and Experimental Medicine (IKE)
University of Health, Linköping, Sweden
Doctorial theses: On Aniridia in Sweden and Norway, Lund University, Lund, Sweden 2009

Epidemiology and eight years follow-up study of Scandinavian aniridia patients

Ulla Edén MD, PhD*
Ruth Riise, MD, PhD**
Kristina Tornquist, MD, PhD***
Neil Lagali PhD*
Per Fagerholm MD, PhD*
* Department of Ophthalmology, Institute for Clinical and Experimental Medicine Faculty of Health Sciences, Linköping University, 581 83 Linköping, Sweden
** Department of Medical Genetics, Rikshospitalet, University of Oslo, Norway
***Department of Ophthalmology, Lund University, Sweden
Corresponding author: Ulla Edén

Epidemiology and eight years follow-up study of Scandinavian aniridia patients

Abstract
Purpose. To find out the prevalence of aniridia, document corneal morphology and changes over time in patients with congenital aniridia.

Material. A thorough search for patients with aniridia has been performed in Sweden and Norway 2004-2005. Out of 181 patients 124 were examined. Sixteen of the Norwegian patients have been involved in a follow up study 2012.

Methods. In the first study the examination included test of visual acuity, corneal sensitivity, tear quantity and quality. Slit lamp examination and ophthalmoscopy were supplemented by digital photographs. The second study also included anterior segment optical coherence tomography (ASOCT) and in-vivo confocal microscopy (IVCM).

Results. Epidemiology: The prevalence in the entire region was 1:72 000. In Sweden the prevalence for patients <80 years of age was 1:70 000 and in patients < 20 years 1:47 000. Eight years follow-up study: Thirty two eyes were examined twice. Most of the patients had cataract or had undergone cataract surgery. Keratopathy was divided into stage 1 (the central cornea not involved) and stage 2 (marked central keratopathy). Stage 1 was found in 50% of the eyes. All of those eyes had a normal corneal sensitivity and BUT was only decreased in two eyes (one patient). All eyes with keratopathy stage 2 had a decreased BUT and 50 % also

had a decreased corneal sensitivity. Out of the eyes with keratopathy stage 2 nine eyes had progressed from stage 1.

Conclusions. These studies show that aniridia seems to be more common than previously estimated, and that some complications appear early in life.
Polar lens opacifications were common and cataract develops very early in life. Factors that may predict the progression of keratopathy are decreased tear break up time, corneal sensitivity and ocular surface disorders. The influence of cataract and/or glaucoma surgery on aniridic keratopathy will be reported.

Mahinda Yogarajah, MD, PhD

Dr Yogarajah is a trainee neurologist currently based at the National Hospital for Neurology and Neurosurgery in London. His PhD was in the application of structural and functional imaging techniques to patients with temporal lobe epilepsy. He has an interest in the synergy between neurogenetics and neuroimaging, and is currently working with patients with aniridia in a project led by Prof. Sanjay Sisodiya at the Institute of Neurology, London.

PAX6 and the brain

Abstract

The brain and the eyes share several characteristics including several underlying gene regulatory networks that direct the development of these related, but structurally non-homologous organs. With the advent of novel imaging techniques our ability to characterise brain structure and function is improving. The aim of this talk is to review previous and current work based on magnetic resonance imaging (MRI) techniques that are starting to improve our understanding on the involvement of PAX6 in brain structure and function in humans.

Dr Doris-Eva Bamiou, MD MSc (distinction) PhD

Dr Bamiou is a Clinical Senior Lecturer at the UCL Ear Institute, and Consultant in Audiological Medicine at the National Hospital for Neurology and Great Ormond Street Hospital. She holds a personal Senior Lecturership award by the Department of Health Higher Education Funding Council for England. She trained in ENT in Greece, in Audiological Medicine in the UK and undertook a three-month funded fellowship to train in auditory processing disorders in Professor Musiek's department in USA where she trained. Her PhD is on auditory processing in patients with structural brain lesions.

Dr Bamiou has been Director and Organiser of the Current Trends in Auditory Processing Disorders instructional courses for the past 10 years and Programme Director of the MSc in Audiovestibular Medicine (UCL) since 2010. She has been Secretary elect of the British Society of Audiology, past Chair of the Auditory Processing Disorders Specialist Interest Group (BSA) and Editor of the Neuro-otology Module of the eBrain e-learning module (RCP and EFNS). She is adviser in Audiology to the JLO, and in the Editorial Board of the Audiological Medicine journal. Her research interests include auditory processing disorders in neurological & normal subjects, auditory training, auditory neuropathy, tinnitus, vestibular rehabilitation, balance in the elderly and psychiatric overlap with vestibular disorders.

Auditory processing and *PAX6*

Abstract

Broadly speaking, the term "central auditory processing" refers to how the brain analyses sound to derive meaningful information. Some patient may have hearing and listening difficulties in the presence of normal hearing thresholds due to disordered auditory processing within the brain and this is referred to as an "Auditory Processing Disorder". We found central auditory processing deficits in 6 out of 8 tested adults with PAX6 mutations indicating deficient auditory interhemispheric transfer that could be attributed to the absence/aplasia of the anterior commisure and/or deficiency of the corpus callosum associated with PAX6 mutations. We found similar results in 11 children with PAX6 mutations, who reported significant difficulties with understanding of speech in noise and localization of sounds, in the presence of a normal audiogram and of MRI-documented abnormalities of the interhemispheric pathway. A pilot study of auditory training in 3 children showed some improvement after training. This presentation will discuss auditory processing deficits in individuals with PAX6 mutations and potential management strategies.

DPhil Veronica van Heyningen

Veronica van Heyningen has Cambridge BA in Natural Sciences (Genetics) and an Oxford DPhil degree in cell genetics and early gene mapping. She has worked at the MRC Human Genetics Unit in Edinburgh for 35 years from postdoctoral fellowship to Section Head status until recently. She has worked on the mapping and identification of genes and their analysis in disease. Her work on developmental eye abnormalities led to studies on genetic mechanisms in disease, including ideas on how specific mutations may lead to different phenotypes. Findings in human disease cases also triggered an interest in long-range regulation of gene expression by enhancers and in the function of gene regulatory networks. Variable disease manifestations, even within families, opened up exploration of factors implicated in phenotype modulation by genetic background differences and also in response to variable environments.

Veronica van Heyningen was a founding member of the UK Human Genetics Commission and the recipient of a number of honours including Fellowship of the Royal Society, and of the Royal Society of Edinburgh, as well as of the Academy of Medical Sciences, and Membership of EMBO. She has been the President of the European Society of Human Genetics and the Genetics Society.

Mutational spectrum for aniridia, genotype-phenotype correlation, and lessons for understanding gene function

MRC Human Genetics Unit, Institute of Genetics and Molecular Medicine, University of Edinburgh, UK

Abstract
Classical aniridia is generally caused by the functional loss of one copy of the PAX6 gene, showing that the dosage of PAX6 protein is critical for normal eye development and maintenance. There are many ways to destroy gene function through mutation. All or part of the gene may be deleted, or different mutations may lead to predicted premature protein termination which ultimately leads to no protein being produced from the mutant allele. There is a special class of mutations at the very end of the gene that lead to an abnormally elongated protein which produces a relatively severe retinal phenotype. A minority of PAX6 paired-domain missense mutations lead to aminoacid changes in the protein in a series of non-classical cases with either mild aniridia, or, in a few cases, aminoacid changes may lead to severe disease that clinicians would not normally describe as aniridia. In this category some cases are so severe that they look like mutations in the anophthalmia/microphthalmia gene SOX2.

Thanks to work by a Japanese group, we now know that PAX6 and SOX2 interact at the protein level and they bind together to several targets including a lens protein gene and to upstream regions of PAX6 and SOX2, so there is significant autoregulation in this part of the eye gene network. Carefully regulated gene networks are very important for robust

development and later gene functions. We have also observed that genes like PAX6 can be disrupted by interrupting the DNA outside the coding region of the gene. This observation led to the postulation that genes like PAX6 require a complex regulatory region for correct spatiotemporal gene expression that is necessary for normal eye development. We have explored the mechanisms for regulating gene expression at a distance. PAX6 is just one example of a much more common phenomenon. There are still some mutations that have not been seen in eye disease; perhaps they are associated with a different phenotype.

Dr. J. Lauderdale

Dr. Lauderdale received his doctoral degree from Purdue University in West Lafayette, Indiana, and performed post-doctoral research at the University of Michigan under the direction of Dr. John Kuwada and then Dr. Tom Glaser. He is currently an Associate Professor in the Department of Cellular Biology at the University of Georgia, which is located in Athens, Georgia.

Dr. Lauderdale's research program is directed towards understanding how the human eye is formed and maintained, and the role that the PAX6 gene plays in these processes. His laboratory studies the mechanisms that control how the PAX6 gene is expressed while the eye is developing, and has recently identified a new form of the PAX6 protein that has a role in forming the lens, iris, ciliary body, and cornea. The laboratory is using their knowledge to explore potential cell-based therapies that could be used to treat aniridic keratopathy and glaucoma. Additional interests include understanding the role of PAX6 in brain development and function.

PAX6 and limbal stem cells

Abstract
Aniridia-related keratopathy (ARK) is estimated to occur in >90% of aniridic patients and is an important factor contributing to the progressive loss of vision in these individuals. Although aniridic children start life with clear normal-looking corneas, their corneas begin to change as they age. Corneal changes in ARK include corneal vascularization and conjunctivalization, sub-epithelial fibrosis and stromal scarring, recurrent erosions, corneal ulceration and chronic pain.

ARK is currently managed using limbal stem cell transplantation techniques that are directed at repopulating the aniridic patient's limbal stem cells from donor sources. Techniques that are used include cadaver keratolimbal allograft (KLAL), living related conjunctival limbal allograft (lr-CLAL), and transplanting ex vivo expanded limbal epithelial progenitor cells onto a suitable carrier followed by penetrating keratoplasty. Of these, KLAL and lr-CLAL have been reported for the treatment of ARK, with KLAL the more widely used technique. Although the long-term effectiveness of these transplant techniques is not yet known, the results thus far are encouraging and suggest that limbal transplants can be used as a means of managing ARK in adult patients.

Despite their promise, both KLAL and lr-CLAL have important limitations in the management of ARK. First, both procedures are performed after stromal scarring has progressed to the point that visual function is severely impaired, thus requiring more extensive surgical intervention. Second, both procedures require long-term systemic immunosuppression to prevent rejection of the donor tissue, thus making these procedures less suitable for teenagers, young-adults, and those individuals for whom long-term immunosuppression would be problematic.

Our research is focused on developing a cell-based procedure for the treatment of ARK that uses the aniridic patient's own cells. Our studies on Pax6-deficient mice (Pax6+/-), the murine animal model for aniridia, suggest that ARK may be primarily due to an abnormal corneal epithelial/stromal healing response rather than a loss of limbal stem cells. This idea is consistent with clinical observations in humans and is supported by a recent report in which an elderly aniridic patient successfully underwent repeat penetrating keratoplasty and has maintained a clear cornea for 4 years with continuous bandage lens wear.

We are currently testing if genetically modified corneal progenitor cells can functionally restore the cornea in a rabbit surgical model of limbal stem cell deficiency. Data from these experiments will be presented.

David FitzPatrick MD, PhD

David FitzPatrick is a clinical geneticist and group leader at the MRC Human Genetics Unit. He did his undergraduate training in medicine was in Edinburgh with postgraduate training in paediatrics and clinical genetics in Edinburgh, Bristol, Glasgow and Baltimore. His main research interests are in the genetic basis of human developmental disorders including eye malformations, facial clefts and and syndromal forms of learning disability.

Mutations in or around *PAX6*, *FOXC1* or *PITX2* cause *PAX6*-negative Aniridia

David R FitzPatrick[1], Morad Ansari[1], Helene Dollfus[2], Pierre Bitoun[3], Francoise Meire[4], Brunella Franco[5], Veronica van Heyningen[1]

1. MRC Human Genetics Unit, Institute of Genetics and Molecular Medicine, Western General Hospital, Edinburgh EH4 2XU, UK 2. Service de Génétique Médicale, Hôpital de Haute-Pierre, F-67098 Strasbourg, France 3. Medical Genetics Departments, University Hospital Jean Verdier, Bondy, France 4. Hôpital Universitaire des Enfants Reine Fabiola, Bruxelles, Belgium 5. *Medical Genetics, Department of Pediatrics, Federico II University, 80131 Naples, Italy*

Abstract

Aplasia of the iris can occur as an isolated malformation (classical aniridia) or as a component of rare multisystem disorders. Most classical aniridia is caused by heterozygous loss-of-function mutations at the *PAX6* locus. We studied 26 aniridia cases that had previously scored as negative for intragenic *PAX6* mutations - 10 of these cases were referred with a syndromal entity Gillespie syndrome (GS), characterized by circumpupillary aniridia and cerebellar ataxia. Re-sequencing the PAX6 locus revealed two cases with previously missed heterozygous intragenic *PAX6* mutations. Array-based comparative genomic hybridization (aCGH) identified two whole-gene *PAX6* deletions and a *PAX6* regulatory microdeletion. Identification of a *FOXC1* whole-gene deletion in a GS case with led us to sequence *FOXC1* coding region in the remaining cases identifying two de novo heterozygous missense changes. Another case had a 3.5Mb de novo deletion of 4q25-q26, which removes a putative regulatory region upstream of *PITX2*. Finally mapping of a GS case associated with a de novo t(X;11) translocation disruption of *ARHGAP6* and *PHF21A* by the X and 11 breakpoints respectively. Plausible pathogenic mutations were thus identified in 8/16 aniridia and 3/10 Gillespie syndrome-like cases. These and other data suggest that a dosage sensitive network of transcription factors including *PAX6*, *FOXC1* and *PITX2* is critical for normal development of both the iris and cerebellum.

Morten C. Moe MD PhD:

Morten C. Moe is Associate Professor at Department of Ophthalmology, Oslo University Hospital and University of Oslo. Before he started residency in ophthalmology, Moe was a post-doc at Karolinska Institutet in Sweden working on characterization of adult human stem cells. Moe is heading different research projects related to translational research at the department and is core member of the Norwegian Center for Stem Cell Research.

Ex vivo expanded autologous limbal epithelial cells on amniotic membrane using a culture medium with human serum as single supplement

Abstract

In patients with limbal stem cell deficiency (LSCD), transplantation of expanded human limbal epithelial stem cells can restore the structural and functional integrity of the corneal surface. Even though there is an obvious strive for development of culture conditions with a reduced content of animal products and omission of foreign feeder cell types, their common protocol for expansion before therapeutic use in patients includes cholera toxins, exogenous growth factors, hormones as well as fetal calf serum. We have recently compared human limbal epithelial cells cultivated on human amniotic membranes in a standard complex medium to a medium with autologous human serum as single growth supplement. Furthermore, we have evaluated our first transplantations using this method for patients with LSCD. Our data indicate that a culture medium with human serum as single growth supplement is an equivalent replacement for the commonly used complex medium for *ex vivo* expansion and transplantation of human limbal epithelial cells. Omission of such products may reduce the immunogenicity of the transplanted tissue and also safeguard against the transfer of infectious diseases and therefore ultimately affect the postoperative outcome and the safety of the procedure.

Dr. Elizabeth M. Simpson

Senior Scientist, Centre for Molecular Medicine and Therapeutics (CMMT) Professor, Department of Medical Genetics, University of British Columbia, Vancouver, Canada , Associate Member, Department of Psychiatry, University of British Columbia, Vancouver, Canada

Research Interests: The overall goal of Dr. Simpson's research program is to improve the prevention and treatment of human brain, eye, and behaviour disorders. Hers is a genetic and genomic approach to the investigation of gene expression, stem cells, neurogenesis, and behaviour.
The Simpson laboratory employs multidisciplinary methodologies with a particular focus on generating and studying mouse models of human disease. The expectation is that a clearer understanding of human brain and eye pathologies will translate into new and improved prevention and therapeutic strategies.

Aniridia Gene Therapy: Pre-Clinical Studies

Andrea J. McLeod[1], Russell J. Bonaguro[1], Michelle Zhou[1], Alice Y. Chou[1], Laurie L. Molday[2], Sanford L. Boye[3], Vince A. Chiodo[3], William W. Hauswirth[3], Robert S. Molday[2], Wyeth W. Wasserman[1,4], and Elizabeth M. Simpson[1,4]

[1] Centre for Molecular Medicine and Therapeutics at the Child & Family Research Institute, University of British Columbia, Vancouver, British Columbia, Canada; [2] Department of Biochemistry and Molecular Biology, Centre for Macular Research, University of British Columbia, Vancouver, British Columbia, Canada; [3] Department of Ophthalmology, University of Florida, Gainesville, Florida, USA; and [4] Department of Medical Genetics, University of British Columbia, Vancouver, British Columbia, Canada;

Abstract
Aniridia is a rare dominant genetic eye disorder in which individuals are born with low vision that, despite current therapies, typically progresses to blindness. Approximately 80% of these individuals carry a mutation affecting one copy of the transcription factor *PAX6*. "Gene therapy", or more appropriately, gene-based protein delivery, can be used to deliver a missing gene or protein to cells that need it. The eye is an excellent organ for gene therapy because it is easily accessible. Currently, individuals carrying another rare genetic eye disorder, Leber's congenital amaurosis (LCA), are benefiting from gene therapy. Our long term goal is to make gene therapy an option for patients with aniridia. Our short term goal is to cure the mouse model of aniridia. Curing aniridia in mice will lay the conceptual and practical foundation upon which human gene therapy can be designed.

We are taking a two pronged approach towards gene therapy in aniridic mice. The first objective is to test whether the complete human *PAX6* gene can rescue the entirety of the mouse aniridic phenotype. The second is to test whether adeno-associated viruses (AAV) can ameliorate the mouse aniridic phenotype. In support of both of these objectives, our bioinformatic team has initiated a *PAX6*-regulatory-region database. We have also initiated construction of a single human BAC carrying the entirety of the human gene, including all its known regulatory regions. Finally, we have cloned 3xFLAG/*PAX6* under the control of the smCBA ubiquitous promoter in an AAV2 viral backbone. Virus carrying this construct has been introduced subretinally, and intravitreously into wild-type mouse eyes. By this means we have successful delivered the *PAX6* protein into multiple cell types of the retina.
These experiments are the first towards AAV-based gene therapy for aniridia in mouse.

Liv Drolsum, MD, PhD.

Liv Drolsum is the head of Anterior Segment Section, Department of Ophthalmology, Oslo University Hospital, Ullevål since 2005. Eye surgeon of the anterior segment, with special interest and knowledge in corneal and cataract surgery.
Professor at the Department of Ophthalmology, Institute of Clinical Medicine, University of Oslo since 2006.

Research area: Clinical studies in corneal and cataract surgery. Member of Center for Eye Research group, University of Oslo. The group has wide experience in tissue- and cell culture procedures, program for transplantation of *ex vivo* generated ocular tissue, including transplantation of limbal stem cells and translational research projects where isolation and characterization of stem cells from the adult human eye is one of the main strategic research focuses.

Cataract and corneal surgery in aniridia

Abstract
Aim. A presentation of the experience with corneal and lens surgery in patients with aniridia in our department.

Introduction. Almost all structures of the anterior segment of the eye might be involved in aniridia. Corneal clouding may occur, and is for most part related to limbal stem cell deficiency. The patients suffer from recurrent corneal epithelial defects, chronic inflammation and pain. Aniridia is often associated with cataract, and there is an increased risk of fragile anterior capsule and subluxation of the lens either into the anterior chamber or posteriorly. Since the aniridia patients often have symptoms related to the absence of the iris, surgeons should consider iris substitute together with cataract surgery.

Results and Discussion. The different options in cataract surgery will be mentioned, such as aniridia IOLs, aniridia rings, and techniques for management of the subluxated lens. The use of aniridic devices will be shown. Our research group studying *ex vivo* expanded limbal epithelial cells will be mentioned. Transplantation of limbal fragments will be presented. Another case with totally opaque cornea treated with keratoprosthesis will also be shown.

Per Fagerholm, MD, PhD:

Thesis 9/6 1978.Dept of Medical Physics, Karolinska Institutet, Stockholm. "The response of lens cells to trauma". Tutor professors' emeriti Bo Philipson and Bo Lindström.
Postdoc. in Ophthalmology, August 1981- July 1982. (Grant from Fight for Sight.NY.NY.) Massachusetts Eye & Ear Infirmary, Harvard Medical School, Boston, USA.
Docent 1981 Medical Physics, Karolinska Institutet
Docent 1983 Ophthalmology (Dept of Ophthalmology, Karolinska hospital), Karolinska Institutet, Stockholm, Sweden.

Clinical undertakings:
Head of Medical and Surgical Cornea and Ocular surface Service – Dept of Ophthalmology,University Hospital, Linköping.
Head of the cornea bank, Dept of Ophthalmology, University Hospital,Linköping (Established the first Long term Cornea Bank 1987, Dept of Ophthalmology, Karolinska Hospital, Established a cornea Bank at St Eriks Eye Hospital, Stockholm,Sweden 1990 and then at the Dept of Ophthalmology, University Hospital, Linköping 1991
Professor/senior consultant 2000 - :Chairman Research and education, Dept of Ophthalmology, University Hospital Linköping

Professor Fagerholm has been the main tutor for many PhD students and post docs, and he has been used as thesis opponent several times. He has several undertakings in different boards and committees and is an expert member of the committee converting the EU directive on tissues handling to national directives and a member of the steering committee The Swedish Society for Corneal Surgeons.

The surface of the eye in aniridia

FAGERHOLM P*, EDEN U*, LAGALI N*, PAASKE UTHEIM T, CHEN X**, RIISE R*****

- **Dept of Ophthalmology, University Hospital, Linköping*[2]*
- ***Department of Ophthalmology, Oslo University Hospital, Oslo, Norway;*
- ****The Norwegian Association for the Blind and Partially Sighted*

Corresponding author: Per Fagerholm

Abstract
Purpose: To document ocular surface qualities in patients from Norwegian families with congenital aniridia. 27 patients with a mean age of 27.1 years (range 3 – 62 years) were included

Methods: Detailed ophthalmic examinations were conducted in affected and unaffected members. Digital slit lamp photography, anterior segment optical coherence tomography (ASOCT) and in-vivo confocal microscopy (IVCM) examinations were performedbilaterally to document corneal morphology.Shirmer,BUT and sensitivity was assessed. The keratopathy was classified. The corneas and the lenses were photographed in the slit lamp.

Results: Affected family members presented with different stages of aniridic keratopathy, with a corneal appearance varying from totally transparency to opaque and highly vascularised. Increased corneal thickness, particularly those with severe keratopathy, was noted by ASOCT. By IVCM, opaque corneas were characterized by active vessels and dendritic cells were more common in aniridic corneas. In milder keratopathy, pathologic epithelial findings included epithelial pleomorphism, focal opacities, and degrees of limbal epithelial crypt obliteration were found. Nerves of the anterior cornea exhibited several distinct features, including an unusually close association of subbasal nerves with epithelial cells, an unusually high subbasal nerve density in some of the corneas.BUT was generally shortened in the aniridic group whereas Shirmers test was normal.

Conclusion: Altered epithelial morphology and a vigorous innervation of the anterior cornea werethe most pronounced corneal findings in family members with milder forms of aniridic keratopathy. Further findings confirmed the known increase in corneal thickness and limbal stemcell abnormality in aniridia.In this younger cohort, no distinct association between surgery and activated keratopathy could be seen.

John M. Freeman, MD, PhD

John M. Freeman practices ophthalmology in Memphis, TN, USA in private practice at Memphis Eye and Cataract Associates and as a clinical instructor for the University of Tennessee, Department of Ophthalmology. He received his corneal fellowship under Dr. Edward Holland in Cincinnati, where he received training in ocular surface reconstruction and gained extensive experience treating aniridic patients. Dr. Freeman is medical director for the National Eye Bank Center for Tissues Bank International in Memphis. Prior to medical school, Dr. Freeman received a Masters' Degree in Cellular and Molecular Biology at University of Memphis and as a medical professional has developed a particular interest in the molecular mechanisms behind aniridic keratopathy.

Aniridic Keratopathy: causes and treatments

Abstract

Aniridic keratopathy, a pax 6 gene related condition, involves a slow progressive opacification of the cornea. Typically aniridics are born with clear corneas and then will slowly develop the condition over the first three decades of their life. The keratopathy usually does not decrease baseline vision until the second or third decade of life. The opacification of the cornea begins as a failure of the epithelial layer of the cornea and can progress to cause opacity in the deeper layers of the corneal stroma. Initially, the condition was understood to result from a failure of the limbal stem cells that provide the main reservoir for the continually renewing corneal epithelium. However, basic science studies within the last ten years have made it increasingly clear that mechanisms involving other aspects of corneal epithelial cell maintenance are likely involved in aniridic keratopathy. Current treatments for aniridic keratopathy are largely surgical and are utilized after the condition has progressed to point of vision loss. The two main alternatives for surgical restoration of a clear visual axis are 1) keratolimbal allografts and 2) keratoprostheses. These procedures while largely successful do carry known risks and their long-term success is uncertain. The advent of a true cellular and molecular understanding of aniridic keratopathy and its slowly progressive nature create an opportunity to develop therapeutic strategies that can slow or halt the progression of the keratopathy before surgical intervention is required.

Peter A. Netland, MD, PhD

Peter A. Netland, MD, PhD, DuPont Guerry III Professor and Chair, Department of Ophthalmology, University of Virginia School of Medicine, Charlottesville, Virginia. Dr. Netland received his undergraduate degree at Princeton University, his PhD from Harvard University, and his medical degree from the University of California, San Francisco. He completed his residency in Ophthalmology, followed by a clinical fellowship in glaucoma, at the Massachusetts Eye and Ear Infirmary and was subsequently appointed Assistant Professor of Ophthalmology and Associate Director of the Glaucoma Service at the Massachusetts Eye and Ear Infirmary, Harvard Medical School. He joined the faculty at the University of Tennessee School of Medicine in Memphis, where he was the Siegal Professor of Ophthalmology, Director of the Glaucoma Service and Vice-Chair for Academic Affairs.

In addition to his recognized expertise in the clinical management and surgical treatment of glaucoma, Dr. Netland is an innovative and prolific investigator. In peer-reviewed literature, he has written more than 300 original scientific articles, book chapters, reviews and published abstracts. He has published five textbooks, most recently the second edition of *Glaucoma Medical Therapy* published by Oxford University Press. Dr. Netland has delivered numerous invited lectures and courses on clinical and surgical management of glaucoma, and on glaucoma research at national and international meetings. He is director of a long-standing clinical glaucoma fellowship training program.

The American Academy of Ophthalmology awarded him its Achievement Award in 2001 and its Senior Achievement Award in 2007, and he has served on numerous AAO Committees. He is a Past President of the Memphis Eye Society and served on their Board of Directors, and was a Board member of the Tennessee Ophthalmological Society. He served as an officer of the Chandler-Grant Society, on the Aniridia Foundation International Board of Directors, and as an Examiner and Special Associate Examiner for the American Board of Ophthalmology. Dr. Netland was elected to the American Ophthalmological Society in 2009. Dr. Netland was appointed the DuPont Guerry III Professor and Chair of the Department of Ophthalmology at the University of Virginia in 2009.

Glaucoma in Aniridia

Abstract

Glaucoma is a potentially vision-threatening problem that is commonly encountered in aniridia patients. This condition may develop at birth, or shortly thereafter. More commonly, however, glaucoma is acquired later in childhood or even young adulthood. In a survey of patients with aniridia, the prevalence of glaucoma was 46%. The average age at diagnosis of glaucoma was 13.6 (median 8.5) years. In subjects with aniridia and glaucoma, 76% were treated with glaucoma medications and the majority (58%) had a history of surgical treatment.

In subjects with glaucoma, the mean ± SD number of glaucoma medications was 1.8 ± 1.3 and number of surgical procedures was 1.7 ± 2.0. The mechanism of glaucoma varies, and can include chronic angle-closure glaucoma. Medical and surgical treatments for glaucoma are effective. Surgical treatment is individualized to the patient, and can include prophylactic goniotomy, goniotomy, trabeculotomy, trabeculotomy and trabeculectomy, trabeculectomy with mitomycin C, glaucoma drainage implants, and cyclodestructive procedures. If unrecognized and untreated, glaucoma can result in loss of vision. Thus, it is important to be vigilant for this condition in children affected with aniridia.

Kathryn Colby, MD, PhD

Kathryn Colby, MD, PhD, cornea surgeon at Massachusetts Eye and Ear Infirmary in Boston and an Associate Professor of Ophthalmology at Harvard Medical School

Kathryn Colby, MD, PhD is a cornea surgeon at Massachusetts Eye and Ear Infirmary in Boston and an Associate Professor of Ophthalmology at Harvard Medical School. Following undergraduate work at Johns Hopkins and a PhD in Neurobiology at Brown University, she graduated summa cum laude from the University of Maryland Medical School. Dr. Colby completed residency, chief residency and fellowship at Mass Eye and Ear, where she has been on staff since 1996. Dr. Colby's areas of expertise include Fuchs' corneal dystrophy, novel surgical treatments for corneal diseases including the Boston keratoprosthesis, and ocular surface tumors. In addition, Dr. Colby has an active pediatric cornea practice at Children's Hospital Boston. She has a special interest in clinical research and served for many years as the founding director of the Joint Clinical Research Center, a collaborative endeavor between MEEI and the Schepens Eye Research Institute, Boston, as well as chair of the MEEI IRB. Dr. Colby has numerous publications and is invited to speak nationally and internationally on both corneal and clinical research topics. She has served on multiple committees for the American Academy of Ophthalmology and is currently the Chair of the Cornea Subcommittee of the Annual Meeting Planning Committee. She was recently elected to the Board of Directors of the Cornea Society.

The Boston Keratoprosthesis for Aniridia: Avoiding Complications and Optimizing Outcomes

Abstract
The success of traditional corneal transplantation in patients with aniridia is limited by the lack of healthy corneal epithelial stem cells in this disease. The Boston keratoprosthesis (Boston KPro) is safe and effective for visual rehabilitation for aniridia-associated corneal disease and does not require immunosuppression. Using a case-based approach focusing on aniridia, this talk will review the history of the Boston KPro, the pre-operative evaluation of patients considering Boston KPro surgery, and surgical and post-operative strategies to optimize outcomes. Prevention and management of long-term complications will be emphasized.

Submitted abstracts

Speakers Saturday before lunch 1/4: Tor Paaske Utheim

Treating limbal stem cell deficiency without applying limbal stem cells. What are the options?

Tor Paaske Utheim[1,2,3], Øygunn Aass Utheim[1,2], Jon Roger Eidet[1,3], Sten Ræder[4,5], Xiangjun Chen[3], Maria de la Paz[6], Edward Messelt[7], Borghild Roald[8], Darlene Dartt[9], and Torstein Lyberg[1]

[1]*Department of Medical Biochemistry, Oslo University Hospital, Oslo, Norway*
[2]*Department of Ophthalmology, University of Oslo, Oslo University Hospital, Norway*
[3]*SynsLaser Kirurgi, Oslo/Tromsø, Norway*
[4]*Department of Ophthalmology, Stavanger University Hospital, Stavanger, Norway*
[5]*Department of Clinical Medicine, Section of Ophthalmology, University of Bergen, Bergen, Norway*
[6]*El centro de Oftalmología Barraquer, Barcelona, Spain*
[7]*Institute for Oral Biology, Dental faculty, University of Oslo, Norway*
[8]*Department of Pathology, Oslo University Hospital, University of Oslo, Norway*
[9]*Schepens Eye Research Institute, Harvard Medical School, Boston, MA*
Corresponding author: Tor Paaske Utheim

Treating limbal stem cell deficiency without applying limbal stem cells. What are the options?

Abstract
A wide array of cell-based strategies to treat limbal stem cell deficiency (LSCD) have been proposed over the past 62 years, including amniotic membrane, representing the first cell-based procedure in 1940; transplantation of conjunctival-limbal-corneal epithelium, marking the first limbal stem cell transplantation in 1965; and non-cell based approaches, such as oxygen and steroid pulse therapy. The first transplantation applying cultured cells dates back to 1997. At that time, limbal stem cells were used. However, in order to treat bilateral LSCD, which is more common than unilateral disease, with autologous sources a number of cell types have been proposed during the past decade. These cells offer great potential as they circumvent issues related to allografts, such as transmission of microorganisms and graft rejection. Past, present, and possible future treatment options are discussed.

Speakers Saturday before lunch 2/4: Tor Paaske Utheim

Storage and transportation of cultured limbal and conjunctival epithelial cells for ocular surface reconstruction

Tor Paaske Utheim[1,2,3], Øygunn Aass Utheim[1,2], Jon Roger Eidet[1,3], Sten Ræder[4,5], Xiangjun Chen[3], Maria de la Paz[6], Edward Messelt[7], Borghild Roald[8], Darlene Dartt[9], and Torstein Lyberg[1]

[1]*Department of Medical Biochemistry, Oslo University Hospital, Oslo, Norway*
[2]*Department of Ophthalmology, University of Oslo, Oslo University Hospital, Norway*
[3]*SynsLaser Kirurgi, Oslo/Tromsø, Norway*
[4]*Department of Ophthalmology, Stavanger University Hospital, Stavanger, Norway*
[5]*Department of Clinical Medicine, Section of Ophthalmology, University of Bergen, Bergen, Norway*
[6]*El centro de Oftalmología Barraquer, Barcelona, Spain*
[7]*Institute for Oral Biology, Dental faculty, University of Oslo, Norway*
[8]*Department of Pathology, Oslo University Hospital, University of Oslo, Norway*
[9]*Schepens Eye Research Institute, Harvard Medical School, Boston, MA*
Corresponding author: Tor Paaske Utheim

Abstract
Transplantation of cultured conjunctival and limbal epithelial cells has emerged as promising techniques for treating severe ocular surface diseases. The culture methods hitherto used vary with respect to preparation of the harvested tissue, choice of culture medium, culture time, culture substrates, and supplementary techniques. The possibility of storing cultured cells for at least some days has several important implications: First, increasingly stricter regulations for cell therapy and more advanced technology are likely to lead to centralization of highly specialized culture laboratories at the expense of smaller units. Centralization might be advantageous if it results in gathering of state-of-the-art equipment and expertise, but it necessitates effective transportation strategies, for which storage technology is mandatory. Therapy with cultured keratinocytes and limbal epithelial cells has been suggested for more widespread use. In the years to come, this may be true for a greater number of stem cell technologies, thus increasing the need for storage- and transportation methods. Second, a storage method is important as it allows time for quality and sterility control of cultured cells. Finally, tissue storage is useful to address scheduling issues due to donor variability and production time for cultured cells. Moreover, as cell cultures may fail at any time during cultivation, the planning of surgery becomes cumbersome without validated storage strategies. Our recent results regarding culture, storage, and transportation of conjunctival and limbal epithelial cells are discussed.

Speakers Saturday before lunch 3/4: Luisa Pinello

Visual outcome and rehabilitation in 12 children with congenital aniridia: a clinical study

Pinello L. ,Mazzarolo M.
Paediatric Low Vision Centre, Department of Paediatrics- Padua University.Italy

Abstract
Purpose: To evaluate the following in children with congenital aniridia: age at first examination ,frequency of family history of aniridia, frequency of ocular and general diseases associated with aniridia and visual outcome.

Methods: A retrospective case review was performed and data were collected, including family history, incidence of associated keratopathy, glaucoma, cataract, macular or optic nerve hypoplasia and poor vision. 18 cases were evaluated and 12 of them were followed up.

Results: The mean age at first evaluation was 5.6 years , with 61% sporadic and a 39% familial cases and 2 patients with systemic abnormalities :1 case with WAGR syndrome (Wilms' tumor, aniridia, genitourinary anomalies, and mental retardation) and 1 case with microcephaly and microphthalmia. 6 children had keratopathy (46%), all of them had foveal hypoplasia (100%). 5 patients developed glaucoma (38%) and 9 cataract (69%). Developmental delay was reported in 2 case . At last follow-up the best-corrected visual acuity (BCVA) ranged from 7/10 to light perception (3/10 or more in 38 %) and was count fingers or worse in 6 eyes (23%). A total of 69 % of patients had congenital cataract, 38% glaucoma, 61,5% nystagmus, 46% corneal opacifications.
The best-corrected visual acuity (BCVA) ranged from 7/10 to light perception and was count fingers or worse in 6 eyes (23%).Aniridia was treated with spectacle correction of refractive errors, tinted or photochromic lenses to reduce light sensitivity, occlusion therapy for amblyopia, and low-vision aids such as closed-circuit television and adaptive technology. Cataract extraction was performed in 3 cases. Glaucoma was treated with topical anti-glaucoma medication. Corneal disease was treated with lubricants.

Conclusions: In this study, congenital aniridia was associated with cataract, glaucoma, and keratopathy: an early diagnosis, a careful treatment of complications and a specific rehabilitation approach can improve visual functional outcome.

Speakers Saturday before lunch 4/4: Marco Mazza

Congenital aniridia. Implantation of artificial iris combined with cataract surgery in pediatric age

M. Mazza; A. Del Longo; E. Piozzi
Pediatric Ophthalmology Department, Niguarda Cà Granda Hospital, Milan, Italy.

Abstract
Purpose: To describe the technique and to determine the effectiveness of surgical implantation of artificial iris in pediatric patient with congenital aniridia and cataract.

Methods: A single patient affected by cataract and congenital aniridia was operated on in both eyes with phacoaspiration, IOL and prosthetic iris implantation in the capsular bag. Intraoperative problems, postoperative anatomical outcomes, subjective glare reduction and visual acuity were evaluated.

Results: The patient had improvement of visual comfort with reduction of glare and a gain of visual acuity. There were no intraoperative or postoperative complications.

Conclusions: Cataract surgery in pediatric patients is a very difficult procedure. It is a constant challenge: anatomical features of the great heterogeneity and the particular characteristics of the capsule require caution to avoid changing the surgical plan. In these cases cataract surgery becomes an opportunity to solve some problems arising from congenital aniridia. The insertion of the artificial iris in the capsular bag is the procedure most at risk of complications.

Speakers Saturday after break 1/4: Meeta Pathak

Centre for Eye Research, Oslo University Hospital

Aniridia and limbal stem cell deficiency

Abstract
A healthy cornea is essential for optimal vision. Many aniridia patients (45%) have corneal abnormalities, collectively termed aniridic keratopathy. Aniridic keratopathy is thought to be a result of deficiency of stem cells in the limbus, the transition zone between the cornea and the bulbar conjunctiva. These limbal stem cells are the source of continuous renewal and repair of the cornea. In aniridic keratopathy, the limbal area is partially or totally damaged, resulting in limbal stem cell deficiency (LSCD). Depending on the severity of the disease, aniridia patients with LSCD may experience pain, photophobia and foreign body sensation due to recurrent epithelial defects. Furthermore, ingrowth of conjunctival epithelium and scarring leads to corneal opacity and reduced vision.

Transplantation of ex vivo expanded limbus derived cells is a treatment modality that can restore a healthy and transparent corneal epithelium in patients with LSCD. In unilateral cases, a biopsy is taken from healthy limbus and expanded on amniotic membrane (AM) using culture medium. Cells are cultured on AM for 2-3 weeks. Then, the AM with expanded limbal epithelial cells (LECs) is transplanted to the diseased eye. In aniridia, patients have bilateral and total absence of healthy limbal tissue. In these patients, allogenic LECs from a cadaveric or living relative donor can be used; this requires long-term postoperative immunosuppression. Alternatively, epithelial cells can be obtained from the conjunctiva or the oral mucosa. Presently, our clinic is the only centre in Scandinavia that offers transplantation of ex vivo expanded LECs. At our centre, autologous LECs are cultured in medium with autologous serum as the only growth supplement, devoid of any animal-derived products. Since November 2010, we have treated 13 patients with this transplantation method, including one patient with aniridia.

Speakers Saturday after break 2/4: Marlies Weyns

Allogenic cultivated limbal stem cell transplantation in congenital bilateral aniridia

Weyns, Marlies MD[1], Leysen, Inge MD[1], Zakaria, Nadia MD[1,2], Koppen, Carina MD[1,2], Tassignon, Marie-José MD, PhD[1,2]
[1]Antwerp University Hospital, Department of Ophthalmology, Wilrijkstraat 10, 2650 Edegem, Belgium
[2]University of Antwerp, Faculty of Medicine and Health Sciences, Universiteitsplein 1, 2610 Wilrijk, Belgium

Abstract
Purpose: In congenital aniridia limbal stem cell deficiency is a major cause of visual impairment. In the early stage of the disease, limbal stem cell deficiency can cause chronic inflammation with fibrovascular pannus formation onto the cornea.

Methods: An allogenic limbal stem cell transplantation was performed in two patients with congenital aniridia (mean age 29y). In each case, a 60% HLA-matched parent consented to donate limbal tissue. The limbal stem cells were cultivated on an amniotic membrane for 2 weeks and then transplanted onto the diseased eye after pannus dissection. Systemic immunosuppression was used for one year.

Results: In both cases surgery went uneventful. Patients were monitored closely postoperatively. Increased intraocular pressure was treated accordingly. Soft bandage contact lens wear was continued to prevent epithelial defects. There were no signs of graft rejection postoperatively. In the first case, 12 months after stem cell transplantation, a penetrating keratoplasty was performed. The follow up period was 27 months and the central cornea remained clear. Because of post-keratoplasty astigmatism, a scleral lens was fitted. Patients satisfaction was high since photophobia, pain, visual acuity and quality of vision have improved after stem cell transplantation. In the second case follow up period was 16 months. In this case the central cornea remained opake but there was no new vessel ingrowth onto the cornea. The patient did not experience any improvement in symptoms or quality of vision. Despite the fact that a penetrating keratoplasty could improve the outcome, the patient prefers not to continue further surgical treatment.

Conclusion: Allogenic cultivated limbal stem cell transplantation can be a valid treatment option for limbal stem cell deficiency in aniridia. It can improve symptoms, quality of vision and quality of life.

Speakers Saturday after break 3/4: Bridget C. Ryan

Bioinformatics and Functional Analysis of Predicted MicroRNA. Target Sites in the Mouse Pax6 3'UTR

Bridget C. Ryan[1], Robert Chow, Lauren Braun, Connor O'Sullivan, Perry Howard
Department of Biology, University of Victoria, Victoria, British Columbia, Canada V8W 3N5

Abstract
MicroRNAs (miRNAs) represent a large class of small, non-coding, regulatory RNAs that negatively regulate protein expression via complementary base pairing to mRNA 3' untranslated regions (3'UTRs). They primarily act to fine-tune target gene expression, and play diverse roles during the development and maintenance of multicellular organisms. Notably, some miRNAs regulate proliferation, differentiation, and cell fate choice, while others provide stability and robustness to gene regulatory networks during development. Given the importance of maintaining tight control of Pax6 gene dose during progenitor proliferation and differentiation, and the involvement of Pax6 in developmental gene regulatory networks, we hypothesize that Pax6 expression is directly regulated by miRNAs during vertebrate development.

Here, we use a bioinformatics-based approach to identify predicted miRNA target sites in the mouse *Pax6* 3'UTR, develop a strategy to mutate all prospective target sites, and generate fluorescent reporter constructs having either the wild type or mutated mouse *Pax6* 3'UTR following the coding sequence of green or red fluorescent protein. Our results demonstrate that the predicted mouse Pax6 3'UTR contains 21 highly conserved prospective miRNA target sites, and we identify 39 candidate miRNAs predicted to target the 3'UTR. Of these miRNAs, many are known to be expressed in tissues that also express Pax6, those being: the eye, retina, CNS, pancreas, and olfactory system. Of particular interest are miR-7, miR-9, miR-375, miR-200b/c, miR-96, and miR-182, all of which are known to be involved in developmental processes that also require Pax6. We are now testing reporter plasmid expression in HEK293T cells and retinal explants, where we predict that fluorescence intensity of the wild type Pax6 3'UTR reporter will be decreased relative to the mutant. Our aim is to develop an increased understanding of the mechanisms involved in regulating Pax6 expression with the goal of supporting clinical treatments for Pax6 related diseases like aniridia.

Speakers Saturday after break 4/4: Cheryl Y. Gregory-Evans

Molecular and phenotypic defects caused by *Pax6* mutation in the mouse are corrected by small molecule translational bypass therapy.

Authors: Xia Wang, Kevin Gregory-Evans, Cheryl Y. Gregory-Evans.
Department of Ophthalmology and Visual Sciences, University of British Columbia, Vancouver, BC, Canada

Abstract

Purpose: Aniridia is a bilateral pan-ocular eye disease caused by haploinsufficiency of *PAX6*. The commonest type of mutation in *PAX6* is nonsense mutations causing a premature stop in protein translation. In this study we tested the efficacy of the small molecule drug therapy aimed at bypassing stop mutations to ameliorate the disease phenotype of the $Pax6^{Sey\text{-}Neu}$ mouse model.

Methods: $Pax6^{Sey\text{-}Neu}$ heterozygous pregnant mice (from E12.5) were treated daily with 6.25 μg/g gentamicin or PTC124 until P21. We also use a postnatal paradigm of daily drug therapy in mouse pups from P4-P21. Eye histology and cell proliferation was analyzed by phospho-histone H3 immunohistochemistry. Gene expression changes in Pax6 downstream targets were analyzed by TaqMan real-time PCR.

Results: In mutant mice, increased cell proliferation in the iris and retina was associated with the *Pax6* eye phenotype. The expression of *FoxC1* and *TGFβ2* were up-regulated whereas *Bmp4* was expression was down-regulated in the mutant eye. Pregnant mice treated with gentamicin rescued the proliferative defect in the retina and iris, and normalized the lens and cornea defects. Furthermore, a similar rescue was observed when the newborn pups were treated. At the molecular level, the expression patterns of the Pax6 downstream targets were returned to normal. Similar results were obtained with Ataluren, an orally bioavailable drug that does not have the ototoxic and nephrotoxic side effects associated with gentamicin.

Conclusions: Translational bypass therapy used in a prenatal or postnatal paradigm rescued the histological and molecular defect observed in the *Pax6* mutant eye. Since *FoxC1*, *Tgfβ2* and *Bmp4* gene expression changes were normalized by drug therapy, this suggests that they are in the same signalling pathway as Pax6 and should be considered as candidate genes in aniridia patients where a *PAX6* mutation has not been identified. Using a targeted therapy to overcome the underlying *Pax6* genetic defect is the first evidence that a drug treatment strategy is possible for aniridia.

Panel speakers Sunday morning

Kathryn Colby, MD, PhD

The Boston Keratoprosthesis for Aniridia

Aniridia affects the cornea (the normally-clear "front window" of the eye) and causes the condition called aniridia-associated keratopathy. Patients with aniridia lack the stem cells that produce the healthy surface cells of the cornea. When the stem cells don't work, the cornea becomes hazy and full of new blood vessels, which limits vision. A regular corneal transplant has a poor success rate in aniridia because of the lack of stem cells. One approach to solve this problem is to perform a stem –cell transplant, taking healthy stem cells from a relative or from a cornea donor. However, this approach requires potentially dangerous medications to prevent rejection of the donated stem cells. Another approach, which does not require immunosuppression, is to implant an artificial cornea, called a keratoprosthesis. The Boston Keratoprosthesis, developed at the Massachusetts Eye and Ear Infirmary by Dr Claes Dohlman, can restore vision in patients with aniridia-associated keratopathy. This talk will review the outcomes and potential complications of the Boston Keratoprosthesis in patients with aniridia.

John M. Freeman, MD, PhD

Aniridic keratopathy involves a slow progressive opacification of the normally clear cornea. Most but not all aniridics will develop the condition by the second or third decade of their lives and it can cause significant loss of vision from baseline in its late stages. Current treatments largely are employed after the condition has progressed significantly and are surgical procedures with known but hard to quantify risks. With increasing knowledge about the cellular and molecular mechanisms behind aniridic keratopathy there is hope that therapies will be developed to slow or halt the progression of the condition and eliminate the need for surgical intervention.

Professor Irene Gottlob

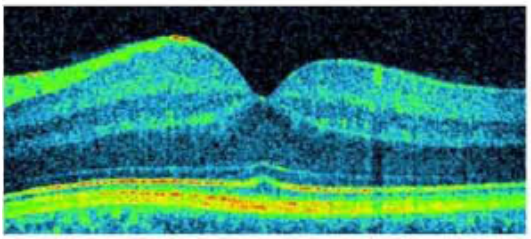
Foveal morphology and eye movements in people with *PAX6* mutations

People with mutation in the PAX6 gene have a variety of changes in the eyes. One of the problems causing reduced vision is that the central retina (fovea), that is needed for sharp vision, does not fully develop (foveal hypoplasia). A new method in ophthalmology, called optical coherence tomography (OCT) allows us to see all the retinal layers, at an almost microscopic level, without even touching the examined person. With OCT we can see how well the central retina is developed and have found that the central retinal development varies widely in different people with PAX6 mutations. We have found that the lengths of the photoreceptor cells (one of the retinal layers) correlate best with the level of vision in foveal hypoplasia and have developed a grading system which can be used to estimate the vision of people with foveal hypoplasia.

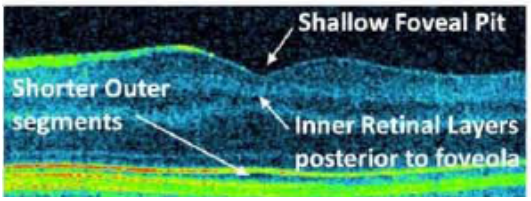
Another problem many people with PAX6 mutations have is nystagmus; an involuntary to and fro or up and down eye movement. We have measured the nystagmus in people with PAX6 mutations and found that it is very variable even within the same families. It is more frequently vertical than in other diseases with foveal hypoplasia for example albinism.

We have recently acquired a hand-held OCT instrument which can be used in infants and small children. We are currently undertaking research to see whether this can predict the visual level children develop later in life.

If you have questions or are interested in participating in our studies in Leicester please contact Irene Gottlob per phone +441162586291 or email (ig15@le.ac.uk).



Normal OCT Tomogram for Comparison



Foveal Hypoplasia Associated with PAX6 Mutations

Dr. J. Lauderdale

Genetics of Aniridia

Aniridia is a rare genetic eye disorder that occurs in about 1 in 60,000 live births worldwide and is typically caused by heterozygous mutations within the PAX6 gene. Approximately two-thirds of aniridic patients have mutations in the portion of the PAX6 gene that encodes for PAX6 protein. The PAX6 protein is a highly conserved transcription factor crucial for normal eye development and cornea homeostasis. An additional 15-20% of aniridic patients have a deletion of the PAX6 gene or a change that prevents the gene from being expressed. The remaining cases of aniridia are caused by mutations in other genes. Understanding the specific mutation is helpful to patients and physicians in managing aniridia.

Peter A. Netland, MD, PhD

Glaucoma in Aniridia

Glaucoma is a potentially vision-threatening problem that is commonly encountered in aniridia patients. This condition may develop at birth, or shortly thereafter. More commonly, however, glaucoma is acquired later in childhood or even young adulthood. In a survey of patients with aniridia, the average age at diagnosis of glaucoma was 13.6 (median 8.5) years. In subjects with aniridia and glaucoma, the majority were treated with glaucoma medications and surgical treatment. Medical and surgical treatments for glaucoma are effective. Surgical treatment is individualized to the patient. The experience of the examination and treatment will be discussed. If unrecognized and untreated, glaucoma can result in loss of vision. Thus, it is important to be vigilant for this condition in children affected with aniridia.

Christopher D. Riemann, M.D.

Aniridia patients present significant challenges to the practicing ophthalmologist both diagnostically and for treatment. The reasons for this as well a rational way forward for diagnosis and treatment will be discussed.

Invited speakers Sunday

Knut Brandsborg

Knut Brandsborg er spesialist i klinisk psykologi. Han har vært ansatt ved Huseby Kompetansesenter siden 1979, med noen år innimellom i barne- og ungdomspsykiatri, voksenpsykiatri og privat praksis. Arbeider i Førskoleteam og Autismeteam, men også en del med skolebarn, ungdom og voksne.

Barns følelsesreaksjoner- hvordan møte dem? Å snakke med barn om deres egen synsnedsettelse

Abstract
Er det noe poeng å snakke med førskolebarn om deres egen synsnedsettelse? Det kan være noen ulemper forbundet med å gjøre det, men fordelene er vanligvis betydelig flere og større. Helt avgjørende vil være måten det gjøres på og at barnet er rede, både i utvikling og i ønsket om å dele noe om dette med en voksen. Den voksne kan fange opp signaler og prøveballonger fra barnet, eller selv prøve å lage åpninger for temaet på forskjellige måter.

Regelmessig samvær med andre som også har en synsnedsettelse kan være et bidrag i seg selv til at et barn får bevisstgjort og bearbeidet tanker og følelser rundt det å være annerledes enn gjennomsnittet.

Presentasjonen er basert på to forutsetninger
- Følelser er alltid sanne, for den som har dem
- Gleder kan bli dobbelt så store og sorger halvparten så store når vi kan få mulighet til å dele dem med andre og de blir tatt i mot

English abstract
Talking with preschool children about their own visual impairment, does it make sense? It may have some negative implications, but there are usually far more advantages. The way we do it is very important. So is the child's readiness when it comes to mental development and his or her wish to share thoughts, feelings and experiences with an adult. The adult may catch signals and "test balloons" from the child, or try to create openings for this theme in various ways.

Repeated contact with other persons who have a visual impairment may in itself contribute to increased consciousness and may also help the child handle thoughts and feelings about being different from the average child.

The presentation is based on two assumptions
- Feelings are always true, for the one who has them
- Joys may be doubled and sorrows be halved when we have the possibility to share them with another person who is able to receive them

Hilde Tvedt Ryen

Utdanning: Allmennlærer, videreutdanning i spesialpedagogikk, synspedagogikk, mobilitetspedagogikk, optometrisk rehabilitering, master i spesialpedagogikk innenfor synspedagogikk.

Erfaring: Syn- og mobilitetspedagog i 14 år, Lærer i grunn- og videregående skole 3 år, Fagleder for spesialundervisning på videregående skole 1 år, Spesialpedagog i forsterket barnehage 4 år, Miljøterapeut for voksen person med autisme 2 år

Yrkestittel: Seniorrådgiver/syn- og mobilitetspedagog

Arbeidssted: Huseby kompetansesenter

Forberedelser til yrkeslivet for personer med synsnedsettelse

Abstract
Kriterier for yrkes- og utdanningsvalg (YOU) kan forankres i motivasjon, langsiktig planlegging, det å foreta realistiske valg og en god porsjon egeninnsats.
Ungdom i dag har mange og varierte YOU. Valgmulighetene er store samtidig som det kan virke overveldende og gjøre det vanskelig å velge.
For å involvere og veilede ungdommen på et tidlig tidspunkt i YOU, ble det i 2008 opprettet et helt nytt obligatorisk fag på ungdomstrinnet som heter «utdanningsvalg». Faget er ment å styrke utdannings- og yrkesveiledning til ungdom i grunnskolen som skal velge utdanningsprogram i videregående skole. Hensikten er å øke elevens valgkompetanse og hindre omvalg og frafall. Grunnlaget for langsiktig planlegging begynner allerede i 8. klasse.

Motivasjon og egen bevissthet om framtidig arbeid er de mest sentrale faktorene for å nå sine yrkesmål. Jeg får ofte spørsmålet «Er det noe han/hun ikke kan bli grunnet nedsatt syn?». Ja, svarer jeg, men det spørsmålet man bør stille: hva er det du kan tenke deg å arbeide med, hva brenner du for, hva er realistisk måloppnåelse for deg? Noen yrker har formelle krav til visus (synstyrke) og synsfelt, men disse utgjør et mindretall av alle yrker.

Det er flere etablerte offentlige ordninger som har som mål å få funksjonshemmede/synshemmede i utdanning og yrke. Uavhengig av de offentlige ordningene, så er det likevel arbeidsgiver som bestemmer hvem som skal ansettes.

Dersom en arbeidsgiver har valget mellom en fullt seende og en som er sterkt svaksynt – hvem er det sannsynlig at arbeidsgiver velger? Vi må også ha arbeidsgivere som våger å ansette personer med synsnedsettelse. Hvilke erfaringer er gjort på dette området?

Preparing for work for people with visual impairment

English abstract
Criteria for regarding occupational and educational choices can be anchored in motivation, long-term planning, evaluate realistic choices and a good portion of self-efforts.

Young people today have a grate range of occupational and educational choices. The options are wide, and it may seem overwhelming and make it difficult to choose.

To involve and supervise youth at an early stage of occupational choices, it was in 2008 created a new compulsory subject in secondary schools called “educational choices”. The course is intended to strengthen the educational and vocational guidance for youth in elementary schools to choose education in high school. The purpose is to increase the student's choice of competence and prevent re-selection and dropout. The basis for long-term planning begins as early as 8th grade at the age of 14.

Motivation and self-awareness are the most important factors to achieve one goals concerning future work. I am often asked, "Is there anything he/she can’t do due to impaired vision?". Yes, I reply, but the question one should ask: what is it you would like to work with, what are your interested in, what is realistic achievements for you? Some professions have formal requirements based on visual acuity and visual field, but these constitute a minority of all professions.

There are established several public services to help disabled/visually impaired in the process of accomplish an education or profession.

Regardless of the public services it is nonetheless an employer who decides who should be appointed. If an employer has a choice between a fully sighted and one that is severely visually impaired - who is likely that the employer chooses? We also need to have employers who dare to hire people with vision loss. What experiences have been made in this area?

Marianne Almbakk

Bachelorgrad i kultur og kommunikasjon fra Institutt for sosiologi og samfunnsgeografi, Universitetet i Oslo.
Mastergrad i psykologi med vekt på kultur og samfunnspsykologi, Psykologisk institutt, Universitetet i Oslo.
Arbeider nå ved institutt for sosiologi og samfunnsgeografi.

Balansekunst: Identitet, selvfølelse og livskvalitet hos ungdom som har en synsnedsettelse

Abstract
Presentasjonen fokuserer på identitet, selvfølelse og livskvalitet hos ungdom som har en synsnedsettelsen og deres relasjoner til foreldre, klassekamerater og venner. Undersøkelsen som denne presentasjonen bygger på har en narrativ psykologisk tilnærming, der deltakernes virkelighet ikke ses på som objektiv, men snarere påvirket av hvem de er, deres tidligere erfaringer og konteksten. Data er semistrukturerte narrative intervjuer som varte i en til to og en halv time, med ni ungdom i alderen 15 til 20 år. Presentasjonen fokuserer på deltakernes egne opplevelser og tanker gjennom sitater fra intervjuene.

Analysen av intervjuene viste at deltakerne beskrev seg selv gjennom aktiviteter de var interessert i og som de mestret godt. Deltakernes rolle i narrativene var en aktiv, flink og sosial person med en travel hverdag. Dette kunne ta oppmerksomheten vekk fra kjennetegn som hadde med synshemningen å gjøre, kamuflere en følelse av ensomhet, og gi deltakerne noe å identifisere seg med. Ungdommene hadde et sterkt ønske om å ha kontakt med andre og føle at de passet inn. Flere framstilte seg imidlertid innledningsvis som sosiale og travle personer, mens fortsettelsen av narrativet viste at de ikke var så mye sammen med venner likevel. Flere av deltakerne hadde en høy terskel for hva de oppfattet som mobbing, og narrativer om vanskelige episoder ble ofte avsluttet med uttrykk som «sånn er livet» eller «det går over». Der deltakerne har et kontaktnett rundt seg, bestående av mennesker som deltar i samme aktivitet eller har samme interesser som dem selv, ser dette ut til å slå ut positivt for deres livskvalitet.

The art of balance: Identity, and social relations of adolescents with visual impairment

English abstract
This presentation focuses on the identity of adolescents with visual impairment and their social relations to parents, classmates and friends. The approach is narrative both concerning theory and method. Data came from semi structured interviews of one to two and a half hours' duration, with nine adolescents between 15 and 20 years of age. The presentation will focus on the participants own experience using quotes to demonstrate the results of the analysis.

The analysis showed that the participants emphasized the importance of being met with realistic expectations. They described themselves in a large degree thru activities they mastered well which in turn seemed to enhance their feelings towards themselves and their social relations. Their wishes for belongingness and social contact with others were strong. Initially, several of the participants constructed themselves as social people with a lot to do, but when continuing, their stories showed that spending time with friends happened quite

infrequently. The participants would not easily characterize episodes as bullying, and narratives about difficult episodes would often end by expressions like “that’s life” or “it will be all right”. Having a network of contacts with the same interests and activity plans, seem to have positive effect on live quality.

Ulla Edén, MD, PhD

Dept. of Ophthalmology, Inst f Clinical and Experimental Medicine (IKE)
University of Health, Linköping, Sweden

Hva skjer med øynene i forbindelse med aldring?

Ulla Edén MD, PhD*, Ruth Riise, MD, PhD**, Kristina Tornquist, MD, PhD***, Neil Lagali PhD*, Per Fagerholm MD, PhD*
* Øyeavdelingen, Institutt for klinisk og ekperimentell medisin,
Linköping universitet, 581 83 Linköping, Sverige
** Avdeling for medisink genetikk, Rikshospitalet, Universitetet i Oslo, Norge ***Øyeavdelingen, Lund universitet, Sverige
Korresponderende forfatter: Ulla Edén

Abstract
Formål: Å øke forståelsen av aniridi.

Materialer: Pasienter i Sverige ble oppsøkt ved hjelp av nasjonalt register over synshemmede barn, synssentraler og øyeklinikker. I Norge ble pasientene kontaktet via den norske aniridiforeningen. 124/181 pasienter ble undersøkt 2004 - 2005. Seksten av de norske pasientene deltok i en oppfølgingsstudie i 2012.

Metoder: Ved den første undersøkelsen ble det gjort en generell synsundersøkelse, supplert med bilder og undersøkelse av tårer. Ved den andre undersøkelsen ble det lagt til moderne metoder for å spesialundersøke hornhinnen.

Resultater: Forekomsten av aniridia i Sverige og Norge til sammen var 1:72 000. I Sverige var forekomsten for pasienter <80 år 1:70 000 og hos pasienter <20 år 1:47 000. Aldersspredningen var fra 6 uker til 79 år. Middelverdien for synsskarphet var 0,2. Vi fant katarakt som påvirket synet hos nesten 50% av pasientene, og glaukom hos 43% av pasientene. Synsnedsettende hornhinneuklarheter ble funnet hos 29% av de undersøkte øynene. Disse forandringene var relatert til alder. I aldersgruppen> 40 år var fordelingen av synsforstyrrende og ikke synsforstyrrende uklartheter omtrent lik. Blant de 10 eldste pasientene hadde åtte pasienter katarakt/var kataraktoperert, syv pasienter hadde glaukom og 8 pasienter (14 øyne) synsforstyrrende hornhinneforandringer.

Konklusjon: De fleste sekundære komplikasjoner ved aniridia er aldersrelatert, men det er betydelige forskjeller mellom pasienter. Mange ulike faktorer bidrar til bl.a. økende hornhinneforandringer. Resultatene av vår oppfølgingsstudie vil bli rapportert.

How does the eye status in aniridia evolve with time?

Ulla Edén MD, PhD[*], Ruth Riise, MD, PhD[**], Kristina Tornquist, MD, PhD[***], Neil Lagali PhD[*], Tor Paaske Utheim MD, PhD[1], Xiangjun Chen PhD[1], Per Fagerholm MD, PhD[*]

[*] Department of Ophthalmology, Institute for Clinical and Experimental Medicine
Faculty of Health Sciences, Linköping University, 581 83 Linköping, Sweden
[**] Department of Medical Genetics, Rikshospitalet, University of Oslo, Norway [***]Department of Ophthalmology, Lund University, Sweden [1] Synslaser AS, Oslo, Norway
Corresponding author: Ulla Edén

English abstract
Purpose: To study the ocular involvement in aniridia and how it evolves over time.

Material: Patients in Sweden were recruited from eye clinics, low vision centers, and from the National Register of Visually Impaired Children. In Norway, patients were recruited with help of the Norwegian Aniridia Society. In total, 124 of 181 patients contacted were examined in 2004-2005. Sixteen of the Norwegian patients participated in a follow-up study in 2012.

Methods: During the initial visit, patients received a general ophthalmic examination including slit lamp photography and examination of tears. During the second visit, a broader examination was completed, with a special focus on detailed examination of the cornea using state-of-the-art ophthalmic instrumentation.

Results: The combined incidence of aniridia in Sweden and Norway was 1 in 72,000. In Sweden, the incidence among those under 80 years of age was 1 in 70,000 and among those under 20 years of age was 1 in 47,000. Aniridia patients in the entire group ranged in age from 6 weeks to 79 years, and the mean visual acuity was 0.2.
Vision-imparing cataract was found in 50% of patients, and glaucoma in 43%. Vision-impairing corneal haze was found in 29%, and was related to age. In those over 40 years of age, the probability of having vision-affecting corneal haze was 50%. Of the 10 eldest patients, 8 had a history of cataract, 7 had glaucoma, and 8 (14 eyes) had vision-impairing corneal haze.

Conclusions: Most complications secondary to aniridia are age-related, but significant differences exist among patients. Additionally, multiple factors appear to contribute to a progressive corneal pathology. The results of the follow-up study in the Norwegian patients will be reported.

Participants

Etternavn	Fornavn	E-post	Stilling/tittel	Firma	Land
Aasly	Karin	karin.aasly@ntnu.no	MD		Norway
Bamiou	Doris-Eva	d.bamiou@ucl.ac.uk	MD, MSc, PhD, DoH, HEFCE Clinical Senior Lecturer & Consultant in Audiovestibular Medicine	UCL Ear Institute, London, UK	United Kingdom
Bobilev	Anastasia	ambobilev@gmail.com	Student	University of Georgia	USA
Bredrup	Cecilie	cecilie.bredrup@helse-bergen.no			Norway
Carstens	Morten Moe	MORTMO@ous-hf.no	MD, PhD Consultant, Department of Ophthalmology, Oslo University Hospital and Assistant professor, University of Oslo, Norway	Department of Ophthalmology, Oslo University Hospital	Norway
Colby	Kathryn	KAColby@MEEI.HARVARD.EDU	Surgeon in Ophthalmology,; Attending Ophthalmologist, Children's Hospital Boston; Associate Professor of Ophthalmology, Harvard Medical School, Boston, MA,	Massachusetts Eye and Ear Infirmary	United States
Drolsum	Liv	liv.drolsum@medisin.uio.no	MD, PhD, Head of section, Department of Ophthalmology, Oslo University Hospital and Professor, University of Oslo, Norway	Department of Ophthalmology, Oslo University Hospital and University of Oslo	Norway
Eden	Ulla	ulla.eden@gmail.com	MD, PhD	University of Linköping	Sweden
Fagerholm	Per	per.fagerholm@liu.se	Professor MD, PhD.	Department of Ophthalmology, University of Linköping	Sweden
Fitzpatrick	Davis R.	david.fitzpatrick@hgu.mrc.ac.uk	MD, FRCP, Professor and Consultant in Clinical Genetics	MRC Human Genetics Unit, Western General Hospital, Edinburgh	United Kingdom
Freeman	John	jmfii@aol.com	M.D., Memphis Eye & Cataract Associates (MECA). Instructor	University of Tennessee Health Science Center Memphis, Tennessee	United States
Gottlob	Irene	ig15@leicester.ac.uk	MD, UnivDoz, FRCOphth Professor of Ophthalmologyr	University of Leicester, Leicester Royal Infirmary, Leicester	United Kingdom

Gregory-Evans	Cheryl	cge30@mail.ubc.ca	Dr	University of British Columbia	Canada
Gregory-Evans	Kevin	kge30@interchange.ubc.ca	Prof	Dept of Ophthalmology, University of British Columbia	Canada
Haugen	Olav	olav.haugen@helse-bergen.no			Norway
Heimdal	Ketil	kheimdal@ous-hf.no			Norway
Husby	Marit Gudrun	maritgud@live.no			Norway
Lauderdale	Jim	jdlauder@uga.edu	PhD, Associate Professor	The University of Georgia, Department of Cellular Biology, Athens, Georgia	United States
Leysen	Inge	inge.leysen@uza.be	Doctor	Antwerp University Hospital	Belgium
Marklund	Lisa	lisa.marklund@jll.se	Low vision specialist	Östersunds sjukhus, Jämtlands läns landsting	Sweden
Mazza	Marco	marco.mazza1@gmail.com	dr		Italy
Mir	Iram Shahzad	i_s_mir75@yahoo.com	MD/Øyelege	Øyeavdelingen OUS, Ullevål	Norway
Moore	Tony	tony.moore@ucl.ac.uk	Professor of Ophthalmology	Moorfields Eye Hospital / Great Ormond Street Hospital, London, UK	United Kingdom
Nerby	Jill	Jill_Nerby@aniridia.net	Founder and Director of Aniridia Foundation International, Congenital Eye Disorder Manager	Department of Ophthalmology, University of Virginia School of Medicine, Charlottesville, Virginia	United States
Netland	Peter	pnetland@gmail.com	MD, PhD, Professor and Chairman of the Department of Ophthalmology	University of Virginia School of Medicine, Charlottesville, Virginia	United States
Oppedal	Truls	truls.oppedal@gmail.com	Dr		Norway
Plinello	Luisa	pinello@pediatria.unipd.it	Prof.	Padua University Paediatrics	Italy
Prescott	Trine	tripre@ous-hf.no	MD, Clinical geneticist	Dept. of medical genetics, Olso University Hospital	Norway
Riemann	Christoper	cbnma@mac.com	M.D., Vitreoretinal Surgeon	Cincinnati Eye Institute,Cincinnati, Ohio	United States
Riise	Ruth	ruthr@online.no		Innland Hospital	Norway
Rustad	Cecilie	cecfr-ru@online.no	MD	Oslo University Hospital	Norway
Ryan	Bridget	ryanb2@uvic.ca		University of Vicotria	Canada
Røsby	Oddveig	oddveig@live.com	overlege	Avd. for medisinsk genetikk	Norway
Simpson	Elizabeth	simpson@cmmt.ubc.ca	PhD, Senior scientist, Professor	Centre for Molecular Medicine and Therapeutics, Department of Medical Genetics, University of British Columbia	United States

Spoz-Zboch	Ewa	etzboch@online.no	øyelege	Øyeavd. Sl Elverum	Norway
Tornqvist	Kristina	kristina.tornqvist@skane.se	Professor	Skanes University Hospita	Sweden
Tønne	Elin	elin.tonne@ous-hf.no		Oslo University hospital, Rikshospitalet, department of medical genetics	Norway
Utheim	Tor Paaske	utheim2@gmail.com	MD PhD	Oslo University Hospital	Norway
van Heyningen	Veronica	veronica.vanheyningen@hgu.mrc.ac.uk	DPhil, FRS, FRSE, FMedSci Professor	MRC Human Genetics Unit, Institute of Genetics and Molecular Medicine, Edinburgh	United Kingdom
von der Lippe	Charlotte	uxhelc@ous-hf.no	MD, specialist in clinical genetics, senior consultant	Oslo University Hospital, Centre for rare disorders	Norway
Vu	Theresa	theresa.k.vu@gmail.com	Dr	Betanien Hospital Skien	Norway
Wankel	Vibeke	vdwankel@online.no	Ophthalmologist		Norway
Wasserman	Wyeth	wyeth@cmmt.ubc.ca	Professor	University of British Columbia	Canada
Wendel	Johan	johanwendel@hotmail.com	lege i spesialisering i oftalmologi	SiV Tønsberg	Norway
Weyns	Marlies	marlies.weyns@uza.be	MD		Belgium
Yogarajah	Mahinda	mahinda2@doctors.org.uk	MD, PhD Clinical research fellow, and specialist Registrar, National Hospital for Neurology and Neurosurgery, Queen Square, London	Institute of Neurology, National Hospital for Neurology and Neurosurgery, Queen Square, London	United Kingdom

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Europeisk medisinsk konferanse i Oslo 8. - 10. juni 2012
Sted: Radisson Blu Scandinavia Hotel i Oslo

Foredrag ved ledende norske og internasjonale eksperter på aniridi. Nyttig for oftalmologer, genetikere og pediatere: Epidemiologi og longitudinale studier. *PAX6*-genet. Glaukom. Retina. Katarakt og korneakirurgi. Genterapi og stamcelleterapi.

For fullt program og påmelding, se www.aniridiaconference.org.
NB! Tellende timer i medisinsk genetikk, oftalmologi og pediatri.

ExtraStiftelsen
Helse og Rehabilitering

ANIRIDI NORGE

Arrangør er foreningen Aniridi Norge.
Konferansen er finansiert med støtte fra ExtraStiftelsen Helse og Rehabilitering og Norges Blindeforbund.
Vår samarbeidspartner er Senter for sjeldne diagnoser (landsdekkende kompetansesenter for cirka 70 sjeldne diagnoser).
Leger ved Oslo Universitetssykehus har faglig ansvar for konferansen.

Learn more about the rare eye disease Aniridia

European Conference on Aniridia, Oslo, Norway, June 8-10th 2012.

The conference fee for medical professionals is NOK 1100 until April 1st. The fee includes meals and accomodation. Late registration fee: NOK 1300.

Venue: Radisson Blu Scandinavia Hotel, Oslo

European and American experts will meet in the capital of Norway to share last findings in aniridia diagnosis and treatment. Presentations will be relevant for ophtalmologists, geneticists and pediatricians:

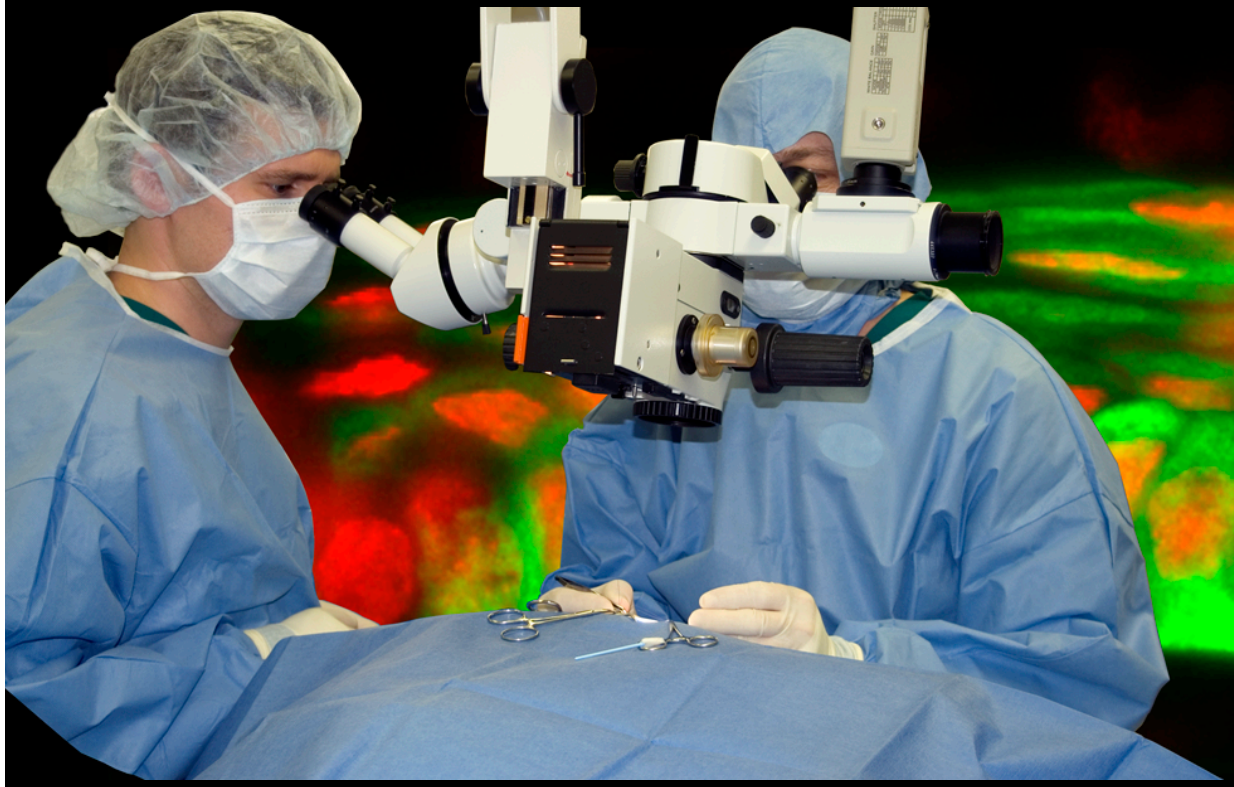
- Epidemiology and longitudinal studies
- *PAX6* genome
- Glaucoma
- Retina
- Cataract and corneal surgery
- Genome therapy and stem cell therapy

For complete program and registration, see www.aniridiaconference.org.

The **European Conference on Aniridia is arranged by the Norwegian Aniridia association with funding from** ExtraStiftelsen and The Norwegian Association of the Blind and Partially Sighted.

ExtraStiftelsen
Helse og Rehabilitering

Genetic Modification and Transplant of Limbal Epithelial Cells

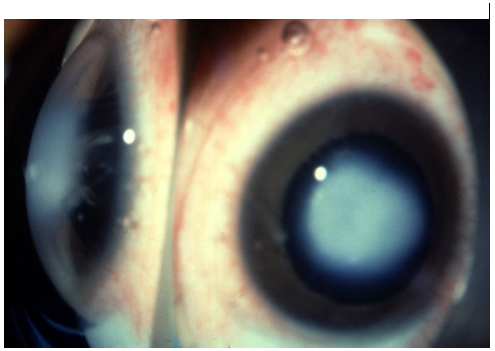


James D. Lauderdale, Ph.D.
P. Anthony Moore, DVM

Departments of Cellular Biology and
Small Animal Medicine and Surgery
The University of Georgia
Athens, Georgia
United States

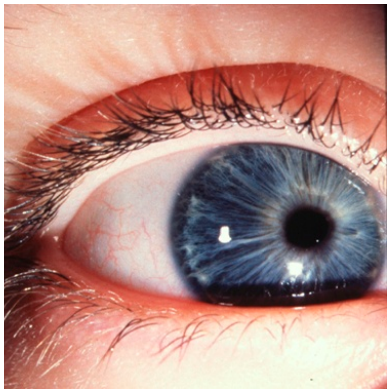
Peters anomaly

R26G



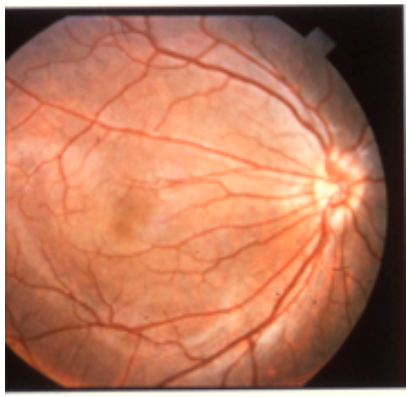
corectopia

V126D



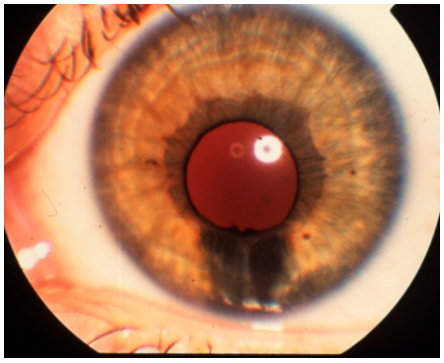
isolated foveal hypoplasia

R128C x 2

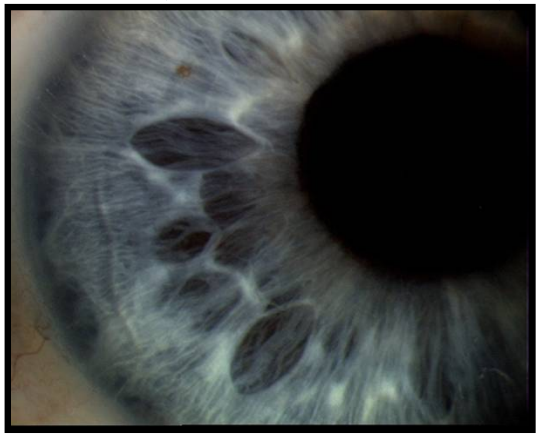


unilateral iris coloboma

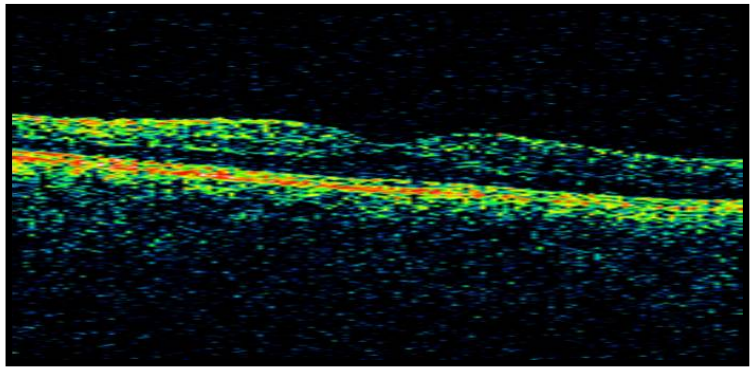
R242T: **HomeoD**



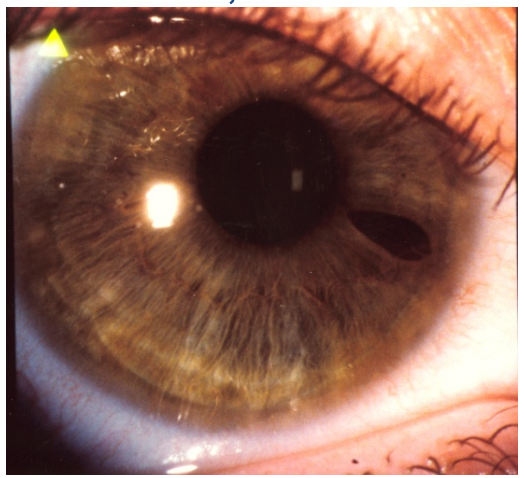
Hanson et al, Nat Genet 6:168-73, 1994; Hum Mol Genet 8: 165-72, 1999; Azuma et al, Nat Genet 13, 141-2, 1996; Morrison et al, J Med Genet 39: 16-22, 2002



G36R – only mild iris anomaly x several



K55R – no foveal hypoplasia



S85S leaky splice mutation

Hingorani et al, IOVS 50: 2581-90, 2009

Pressemelding

Distribusjon: Redaksjonene

Europeisk aniridikonferanse i Oslo

Fredag 8. juni åpner den *første europeiske medisinske konferansen om den sjeldne øyesykdommen aniridi i Oslo.*

Den fremste kompetansen i verden innen oftalmologi (øyeleger) og genetikk er representert. Øyeleger og genetikere fra Norge, øvrige Europa og USA vil i løpet av konferansen presentere den seneste forskningen om aniridi og behandlingen av denne sjeldne tilstanden.

Tid og sted: Konferansen arrangeres på Radisson Blu Scandinavia Hotel i Oslo fra 8. - 10. juni 2012

Noen av de medisinske temaene som belyses under konferansen er

- Epidemiologi og longitudinale studier
- *PAX6-genet*
- Glaukom
- Retina
- Katarakt og korneakirurgi
- Genterapi og stamcelleterapi

Second opinion: Parallelt med den medisinske konferansen arrangeres klinikk på Huseby kompetansesenter. Her kan pasienter få en ”second opinion” fra en av de besøkende legene.

Om aniridi: Medfødt aniridi er en sjelden og alvorlig øyesykdom som medfører synsvansker. Aniridi viser seg som en generell underutvikling av øyet. Typisk mangler iris helt eller delvis. I tillegg er det også økt risiko for andre problemer med øynene. Tilstanden er arvelig, men kan også oppstå spontant. Så langt man kjenner til skyldes aniridi en medfødt forandring av *PAX6-genet*.

I Norge defineres en diagnose som sjelden når det er færre enn 100 kjente individer per million innbyggere i landet. Per 2011 er det registrert cirka 60 personer med aniridi i hele Norge.

Konferansens hjemmeside www.aniridiaconference.org inneholder fullt program med oversikt over foredragsholderne.

Arrangør: Det er interesseorganisasjonen Aniridi Norge (www.aniridi.no) som arrangerer konferansen. Formålet er å fremme forskning og behandling av aniridi. I tillegg er det hensikten at leger og forskere skal få økt kompetanse om øyesykdommen og styrket sitt faglige nettverk.

Presse: Det vil være mulig å arrangere intervjuer med pasienter, leger og representanter for arrangøren, Aniridi Norge.

Kontakt: Ønsker du mer informasjon om konferansen eller undersøke muligheten for å gjøre en intervjuavtale, ta kontakt med informasjonsansvarlig **Bjørn Eckblad (mobil 932 56 205 eller bjorn.eckblad@gmail.com)**

Samarbeidspartnere: Konferansen finansieres av ExtraStiftelsen Helse og Rehabilitering og Norges Blindeforbund. Våre samarbeidspartnere er Senter for sjeldne diagnoser (landsdekkende kompetansesenter for 70 sjeldne diagnoser) Øyeavdelingen og Avdeling for medisinsk genetikk, alle ved Oslo Universitetssykehus, samt Huseby kompetansesenter. Vi samarbeider videre med de europeiske interesseorganisasjonene Aniridi Europe og Aniridia Foundation International.

ANIRIDI NORGE

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Foreningen organiserer ca 7000 overleger ved norske sykehus

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Bjørn Eckblad

Ikke glem øyehelgen i Oslo 8.–10. juni! Vi tilbyr: A) Medisinsk fagkonferanse om øyesykdommen aniridi – www.aniridiaconference.org. B) Konferanseavgift kun kr. 1100 (inkl. måltider + overnatting på SAS-hotellet. Forutsetter påmelding innen 1. april) C) Topp norske og internasjonale foredragsholdere. D) Tellende timer – sjekk kurs her http://bit.ly/GJQjNi. Vi ønsker flest mulig oftalmologer, genetikere og pediatere velkommen.
Hilsen oss i Aniridi Norge.

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1st European Conference on Aniridia

Kursnr.: L-26739

Godkjenninger: Barnesykdommer Videreutdanning: Valgfrie kurs: 12t Etterutdanning: Valgfrie kurs: 12t. Medisinsk genetikk Videreutdanning: Valgfrie kurs: 12t Etterutdanning: Valgfrie kurs: 12t. Øyesykdommer Videreutdanning: Valgfrie kurs: 12t Etterutdanning: Valgfrie kurs: 12t.
Målgruppe: Leger som arbeider med undersøkelse, diagnostikk og behandling av barn og voksne med øyesykdommer.
Læringsmål: Å bidra til bedret komptanse i diagnostikk, behandling og oppfølging av pasienter med aniridi.
Kurskomite: Trine Prescott, overlege og Charlotte von der Lippe, overlege (kursledere), Liv Drolsum, overlege, Ruth Riise, overlege, Bjørn Eckblad, Aniridi Norge, Hilde R. Hansen, Aniridi Norge, Asbjørn Akerli, Aniridi Norge.
Påmelding til: Meeting Management.Meeting Management AS
Niels Juels gate 39
0257 OSLO

Telefon: 22 55 50 11
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Informasjon

Kursnr.: L-26739
Dato:
08.06 2012 08:00-10.06 2012 17:00.
Arrangør: Den norske legeforening

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Sjeldne tilstander | Helsedire

www.helsedirektoratet.no/helse-og-omsorgstjenester/sjeldne-tilstander/Sider/default.aspx

Diagnoser

Kompetansesentre

Nyfødtscreening

Servicetelefonen

Sjeldne tilstander

En medisinsk tilstand regnes som sjelden når det er mindre enn 100 kjente tilfeller per million innbyggere i landet. I Norge tilsvarer dette færre enn 500 personer. I de fleste grupper er det langt færre enn 500 med samme diagnose.

Vi antar at det her i landet er om lag 30 000 mennesker med en medfødt tilstand som det ikke finnes flere enn 100 individer av på én million innbyggere.

Sjeldne tilstander er medfødte og personene har ofte sammensatte funksjonsvansker med behov for tverrfaglige og tverretatlige tiltak. Mennesker med sjeldne og lite kjente diagnoser og funksjonshemninger har, i likhet med andre med nedsatt funksjonsevne, behov for et godt tilrettelagt samfunn.

Helsedirektoratet er faglig forvaltningsorgan for sjeldne tilstander, og har rådgivningsfunksjoner overfor departementene i økonomiske og faglige spørsmål. Direktoratet har ansvar for informasjonstiltak og videreutvikling av feltet.

- Sørge for tilgjengelig kompetanse og kunnskap om sjeldne medisinske tilstander til personer som har slike tilstander, pårørende og personell i tjenesteapparatet
- Bidra til at personer med sjeldne tilstander skal få bistand til å leve et mest mulig selvbestemt liv
- Bidra til å videreutvikle kompetanse- og kunnskapsmiljøer slik at ressursene utnyttes godt

Abonner på RSS-feed

Publikasjoner

Født med en sjelden diagnose. Med sterk vilje og systematisk hjelp kan hverdagen bli bra!

Last ned

En sjelden guide

Last ned

Ekstern konferanse

Oslo 8.-10. juli:

Internasjonal konferanse om den sjeldne øyesykdommen Aniridi