



Clinic for
Special Children®

Newsletter

ISSUE #60

FALL 2023



Ava with her bunny

Turning Insight into Action The Esh Family

When Melvin and Malinda Esh were expecting their first child in 2021, their midwife suggested they get carrier testing to see if they could potentially pass on a genetic condition to their baby. She recommended the couple get tested through the Clinic for Special Children's Plain Insight Panel™ (PIP) - a carrier test that screens for over 1,300 genetic variants specific to the Plain communities of North America (learn more on page 4).

"Our midwife offers the PIP to all of her Plain patients. I knew Glutaric acidemia type 1 (GA-1) ran in my family but we didn't know of anyone in Melvin's family with a rare genetic disorder. We decided to get tested out of curiosity and to be safe," explains Malinda. GA-1 is a rare metabolic disorder that can cause permanent brain injury from the build up of specific amino acids that the body can't process properly.

The PIP results showed that both Malinda and Melvin were carriers for GA-1 and therefore had a 25% chance of their baby having GA-1. "We were very shocked to get the results. As soon as we found out, the Clinic called us to schedule a prenatal visit to provide us with more information about GA-1 and develop a plan for when our baby would be born," said the Esh family. Dr. Grace Meier, Family Medicine Physician at the Clinic, met with the couple at their first visit and shared information about GA-1 and what they could expect.

Several months later, Melvin and Malinda welcomed their first child, a daughter, named Ava. When Ava was born, the midwife took a cord blood sample and drove it to the Clinic for urgent diagnostic testing. Within just several hours after Ava's birth, Melvin and Malinda had an answer -

the test was positive for GA-1.

"When Ava was born and we received her diagnosis of GA-1, we were grateful that we had already developed a relationship with Dr. Meier and providers at the Clinic and had a plan in place. We had special GA-1 dietary formula on-hand to feed Ava to protect her from injury. This was given to us by Dr. Meier at our prenatal consult. The very next morning, we visited the Clinic for Ava's first appointment," says Malinda. "It was already overwhelming becoming new parents, and I can't imagine if we weren't prepared a bit beforehand."

Today, Ava is a thriving two-year-old who enjoys playing outdoors and with the animals. She has been injury-free and is finally allowed to have more natural protein in her diet. "For the past two years whenever Ava had an illness with a fever and had to be hospitalized as a precaution, the doctors from the Clinic treated us amazingly and were there for us. Each provider has treated us with kindness and the Clinic feels like home," says Melvin and Malinda.

"We would highly recommend to anyone considering the PIP test that they get tested. It helps you prepare before your baby is born. If we didn't do the PIP test, we wouldn't have had bottles and formula on-hand, and we would not have found out that Ava had GA-1 until her newborn screen test came back several days after her birth," they explain. "Having the prenatal visit and meeting Dr. Meier before Ava was born allowed us to establish a relationship with our future baby's doctor - even if we didn't realize it at the time! It was helpful when Ava was born and diagnosed that we were working with someone we already knew and met before."



SPECIAL DATES

Finger Lakes Benefit Auction Saturday, October 7

Ontario Produce Auction | Stanley, NY

The Extraordinary Give Friday, November 17

[ExtraGive.org](https://extragive.org)

Online Fundraiser - All Day

Help spread the word about the Clinic during this 24-hour online fundraising event!

Thanksgiving | Office Closed November 23 & 24

Office closed all day

Christmas Eve | Office Closed Friday, December 22

Office closed from 1 p.m. - 5 p.m.

Christmas | Office Closed December 25 & 26

Office closed all day

New Years Eve | Office Closed Friday, December 29

Office closed from 1 p.m. - 5 p.m.

New Years Day | Office Closed Monday, January 1

Office closed all day

Good Friday | Office Closed Friday, March 29

Office closed all day

Events are subject to change due to severe weather conditions, restrictions, or unforeseen circumstances.



Staff News

Joelle Williamson Clark, MPH



Welcome to Joelle Williamson Clark, MPH, who joined the Clinic's team as our first Clinical Research Manager. In this new role, Joelle will be responsible for leading a variety of tasks within research including regulatory submissions and compliance, and study coordination.

Joelle appreciates staying active outdoors hiking, biking, running, and swimming. She enjoys exploring new locations with her husband and their dog, Jarvis, as well as gardening and baking.

Julia Martin



Congratulations to Julia Martin, Development Associate at the Clinic, for celebrating her 5th work anniversary!

In her role, she works with the development team to ensure an excellent experience for donors and stakeholders of the Clinic. Julia has a close connection to the Clinic - as she is the mother of a child living with Maple Syrup Urine Disease (MSUD), who has been cared for by the Clinic.

Erin Sweigert



Welcome to Erin Sweigert, who joined the Clinic recently as a Research Associate. As the Research Associate, Erin will be supporting a number of

the Clinic's research endeavors. These research projects will provide insights into rare genetic conditions and include prospective natural history studies, clinical trials of gene replacement therapies, and the analysis of vital data.

In her free time, Erin enjoys spending

time outside, going to the beach, reading, and spending time with family and friends.

Sarah Thomas, RMA



Welcome to Sarah Thomas, RMA who joined the Clinic's team as a Medical Assistant! In this role, Sarah will assist in providing day-to-day

clinical and administrative services including rooming patients, phlebotomy, cleaning and preparing exam rooms, answering phone calls, and scheduling appointments.

In her free time, Sarah enjoys hiking or going for a walk with her kids.

It's Flu Shot Season!

Please call the Clinic at 717-687-9407 to schedule your or your child's flu shot. Anyone over six months of age is encouraged to get an annual flu shot. We offer flu shots to our patients and their immediate family members. The best way to protect yourself and your family during the flu season is to avoid exposure, practice good handwashing, and receive an annual flu shot.

Clinical Care Charges | Update

The Clinic for Special Children strives to make medical care affordable and accessible for all patient families. As such, the Clinic is now providing clinical care services at no cost to families effective October 1, 2023.

Instead, patient families are invited but not obligated to donate to support the Clinic's mission of providing affordable healthcare for children and adults with genetic disorders or other complex medical needs.

Details about this new model will be provided to families at their appointments after October 1st. We hope that this new model will ensure that the Clinic's clinical care services are affordable and accessible for all families.

Move Update

We are anticipating that our new building will be ready for us to move in around early Spring 2024. All of our operations will be moving to our new building in Gordonville, PA. As we are still about six months away from the Spring, this time frame is subject to change depending on how the rest of construction progresses.

We are planning to send a written letter to all families that have visited the Clinic or used our services when we are about two months away from moving. We will also be communicating move details through social media, newsletters, phone calls, and more. Please stay tuned as we get closer to this exciting move for the Clinic! We look forward to serving your family at our new facility.

Research Update

CouRAGE & research on immune deficiencies at the Clinic

One of the most serious and time-sensitive genetic diagnoses we make at the Clinic is for severe combined immune deficiency (SCID), which can leave a newborn without the immunity needed to fight off common infections, making them very sick soon after birth. We also see that some members of the Plain community have genetic variants that impact their immune function but may not cause serious problems until later in life or following exposure to certain germs. We want to better understand why some people who have these genetic findings experience life-threatening health issues, while others appear to do much better.

As such, we are launching the CouRAGE study this fall, working with families and collaborators around the globe to determine what we can do to best predict- and influence- optimal health outcomes for these patients. We will follow patients with these genetic diagnoses (RAG1 or RAG2-associated immunodeficiencies) and study their immune systems and exposures. These findings may impact how and when we treat these children, and lessons learned will extend beyond these conditions and outside the Plain communities as well.

EXTRATM GIVE



Friday, November 17
ExtraGive.org

It's almost that time of year again! The **Extraordinary Give** - Lancaster County's largest day of online giving - is on **Friday, November 17th**! Help the Clinic **raise \$75,000 in 24 hours** for our mission. You don't need to be located in Lancaster County - we have donors from all over the United States support the Clinic through the ExtraGive.

Every dollar you donate on November 17th during the ExtraGive will be stretched by a pool of more than \$500,000 from local sponsors.

Donating is simple - here are **four ways you can support**:

- 1 On Friday, November 17th (from midnight to 11:59 p.m.) **visit www.ExtraGive.org** and select 'Clinic for Special Children' to give online during the 24-hour period.
- 2 **Call the Clinic at 717-687-9407** between 9:00 a.m. - 5:00 p.m. on Friday, November 17th to donate via credit card over the phone.

Won't have access to internet on the day of ExtraGive? Call us before and we'll help process your ExtraGive donation for you on the day of ExtraGive.

- 3 **Set up your own fundraising page!**

You can create your own personal fundraising page to share with your friends and family to raise money for the Clinic! Visit www.ExtraGive.org, search for 'Clinic for Special Children', and click the 'Fundraise' button to create your own page before November 17th!

Scan the QR code
for a shortcut to
our giving page!



THANK YOU! 2023 Benefit Auctions

We're so grateful to everyone that supported the auctions this year - committee members, attendees, donors, and volunteers! Your support does not go unnoticed. Thanks to you, this year brought a record-breaking auction season. Please enjoy several photos below from this year.



Providing Insight

An update on our Plain Insight Panel™ test

The Plain Insight Panel™, or PIP, is a genetic test developed and only available at the Clinic for Special Children that provides health information associated with 1,300 genetic changes found in Old Order Amish and Mennonite communities. Since the test can identify adult carriers of recessive genetic conditions (disorders that occur when a child receives one non-working copy of a gene from each unaffected parent) it is best if couples both do the PIP so they can learn if any of their children could be born with specific disorders found within the Plain communities. As experienced by the Esh family (see front page), this type of proactive testing before or during pregnancy is a great way to ensure the best start for a baby after birth. Results of the PIP make a difference for carrier couples by:

- informing care during pregnancy and at delivery
- preparing for rapid diagnostic testing and/or treatment for any affected children
- setting the plan and road map for their child's best health as they grow

The PIP can also provide information that can affect the health of the person taking the test. Many of these findings are actionable, meaning that there are ways to minimize their harmful impact(s).

Three-Tiered Insight

The genetic variants included on the PIP are separated into three tiers based on how much we know about the effects they have on health:

- **Tier 1** variants are known to cause disease. However, the severity of the condition depends on the specific variant.
- The majority of variants fall into **Tier 2**. These rare variants are suspected to be disease-causing, but we have not yet identified symptomatic patients who carry them. By following these genetic changes in the population, we can learn more about their contributions to human health.
- **Tier 3** variants are health modifiers. They include variants that are harmless on their own but that can affect health in the presence of certain medications or other genetic variants.

PIP by Numbers

To date, the Clinic's in-house laboratory has completed over 4,500 PIP tests. Of those, the majority of participants (67%) are couples in their childbearing years (18 - 50 years old), though 12% of individuals are between the ages of 50 and 96 years, indicating a multi-generational curiosity in learning about genetic impacts on health. Importantly, over 50% of participants are self-referred or submit samples through a midwife. Physician-ordered tests come internally from the Clinic and from other clinics across the United States.

The average person typically has between 4 and 5 variants and over one-third of couples match for at least one genetic variant. Nearly half of all individuals receive at least one personally-actionable finding. Since launching the PIP in 2019, the Clinic for Special Children has doubled the number of individuals doing the PIP each year and has accordingly increased lab staff and equipment to manage the popularity of this test!

Diagnostic Power of Screening

While most PIPs are performed on adults without specific health concerns for themselves or their children, it is also used by providers seeking a diagnosis for children with developmental or other health issues. In this capacity, the PIP functions as the first-line test to potentially find an answer before moving onto more expensive or time-consuming genetic testing. Roughly 16% of PIPs are done on symptomatic adults or



on parents seeking a diagnosis for a child. If parents match for a condition that fits their child's health issues, we do a targeted test for that condition on the child's DNA to make the diagnosis. The PIP is rarely performed directly on children except in very specific cases and carrier testing is only available to individuals 18 years or older.

New Insights: Reclassifying Tier 2 Variants

We also use the PIP to help us learn more about rare variants that are predicted to affect health and development but have not yet been linked to specific Plain patients with these conditions. By identifying individuals with these Tier 2 variants, studying their health history, and performing follow-up testing, we can learn more about whether the variants are truly harmful or not. If they do not affect health and wellbeing they can then be removed from the PIP.

Tier 2 studies are ongoing, and active participation from the community is critical to providing the most accurate predictors of health. When we find a couple that matches for a Tier 2 variant, we determine next steps together, whether it is testing at-risk family members for the genetic change or doing clinical testing (for example, hearing or vision tests) that further determines the relationship between the variant and disease presentation. Through these studies (performed free of charge), we have reclassified dozens of variants, removing some from the PIP completely and placing others in the Tier 1 category once we identify a patient who has features consistent with the disorder. Altogether, these studies make the PIP a better test for future participants.

What's Next for the PIP?

The Clinic's lab is currently finalizing a third version of the Plain Insight Panel! This updated version will include additional genetic variants associated with new conditions found in Plain communities (see examples in the table below) and many others we suspect impact health. Building a new version of the PIP demonstrates how much we continue to learn about the relationship between genetics and health in the Plain community and also shows how our partnership with the Plain communities fosters overall public health and knowledge.

Gene	Disease	Population
RAG1	Severe combined immunodeficiency	Mennonite
EXOSC3	Pontocerebellar hypoplasia	Amish
TSEN54	Pontocerebellar hypoplasia	Mennonite
KCNQ1	Long QT syndrome (autosomal dominant)	Amish
CABP2	Deafness	Mennonite
POMT1	Muscular dystrophy - dystroglycanopathy type	Mennonite
CFI	Complement factor I deficiency	Amish



The University of Maryland Amish Research Clinic

Collaborator Spotlight

The University of Maryland Amish Research Clinic

Identifying common causes of high cholesterol and Long QT syndrome in the Amish community

In 1995, Dr. Alan Shuldiner founded The University of Maryland Amish Research Clinic (ARC) to study inherited causes of disease in the greater Amish community. With the help of over 7,000 Amish volunteers, the ARC has been able to uncover genetic changes in the community that are known to cause serious health risks. Learning more about how genetic changes run in families and the specific health problems they can cause helps healthcare providers better identify and treat those who need it.

“Each new discovery from the ARC’s research can help improve the health of the Amish community and beyond,” explains Dr. Amber Beitelshes, Professor at the University of Maryland School of Medicine. “The Amish community has some unique benefits to studying genetic causes of disease. Since the Lancaster Amish were founded by a small group of settlers, and it is rare for outsiders to join the community, certain genetic traits have become amplified over time. With large family sizes and detailed family records, the Amish contribute valuable genetic and health information we would not be able to learn otherwise. Sometimes findings from research studies provide immediate benefits to the participants, but often what we learn is used to help improve health in future generations.”

Dr. Beitelshes is currently working on studies focused on two genes: *APOB* (changes in this gene can cause high cholesterol) and *KCNQ1* (changes in this gene can cause disturbances in heart rhythm and heart rate known as Long QT syndrome). “Participants in the study receive free genetic testing specific to the health condition running in their family. For families with hereditary high cholesterol, we test the *APOB* gene. Changes in the *APOB* gene are found in 1 out of every 8 Amish individuals, making it a common cause of high cholesterol in this community,” describes Dr. Beitelshes.

“For families with Long QT syndrome, we test the *KCNQ1* gene. Changes in the *KCNQ1* gene are found in 1 out of every 45 Amish individuals, making it a common cause of Long QT syndrome in the Amish,” continues Dr. Beitelshes. Depending on the condition found in their family, the ARC offers additional testing, free of charge, during a home visit. For example, those with hereditary high cholesterol in their family will have their cholesterol

level checked, while those with a family history of Long QT syndrome will have an EKG performed to detect any abnormal heart rhythms. Individuals identified with the gene change can choose to be connected with a local healthcare provider, who can discuss possible treatment plans and follow-up care based on their results.

The Clinic for Special Children is proud to collaborate with the ARC on these studies. “The Clinic for Special Children offers discounted clinical testing that allows studies like ours to be conducted. Without their support we wouldn’t be able to offer testing to as many people,” explains Dr. Beitelshes. “For this study, we collect samples for genetic testing when we visit the participants in their homes. The Clinic’s lab performs genetic testing on each sample to confirm the gene change in the affected individual, as well as identifying whether or not their family members inherited the gene change. A research team member will discuss the test results with participants so that they can understand what it means for them, including healthcare recommendations and options for treatment moving forward.”

Findings from the ARC’s studies have also informed updates to the Clinic’s Plain Insight Panel™ (PIP) carrier test. The Clinic will soon be adding the *KCNQ1* genetic change to the PIP test to provide more people with further insight into their health. We are grateful to the ARC for their important work in the Amish community!

To learn more:

For community members who are interested in or want to learn more about high cholesterol and Long QT syndrome, healthcare providers who specialize in heart conditions have been offering informal educational sessions. The meetings will be held in Amish homes, and it is a time where people can bring copies of their cholesterol test results and ask questions they may have about their levels and treatment options. These sessions will be free of charge, and details on possible upcoming meetings can be found by calling the Amish Research Clinic at 717-392-4948.

Our Staff

Amy Albright, MS, CGC
Genetic Counselor

Keturah Beiler, BSN, RN, CHPPN
Nurse & Cherished Lives Program Manager

Karla W. Brigatti, MS, CGC
Research Operations Director

Vincent Carson, MD
Pediatric Neurologist

Joelle Williamson Clark, MPH
Clinical Research Manager

Kelly Cullen
Marketing and Communications Manager

Skye Gawn
Gift Processing and Data Entry Coordinator

Jennifer Giacoio
Medical Receptionist

Adam D. Heaps, MS, MBA
Executive Director

Christine Hendrickson, RNC
Nurse

Candace Kendig, RMA
Office Manager

Alanna Koehler, PhD
Assistant Laboratory Director

Lavina King
Community Liaison

KaLynn Loeven
Laboratory Scientist

Julia Martin
Development Associate

Alexis McVey, BSN, RN, CPN
Nurse Manager

Grace Meier, MD
Family Medicine Physician

Laura Poskitt, DO
Pediatrician

Erik G. Puffenberger, PhD
Laboratory Director

Stephen D. Ratcliffe, MD, MSPH
Senior Consulting Physician

Donna L. Robinson, MSN, CRNP
Nurse Practitioner

Ashlin Rodrigues, MS
Laboratory Scientist

Angela M. Sepela, MBA
Development Director

Dawn Sheets
Medical Receptionist

Jessica Snyder, PHR, SHRM-CP
Human Resources Generalist

Kevin A. Strauss, MD
Medical Director

Erin Sweigert
Research Associate

Sarah Thomas, RMA
Medical Assistant

William Van Ess, MS, CFE
Accounting Manager

Susan Walther, MS, CGC
Genetic Counselor

Kelly Woodson
Event and Volunteer Manager

Board of Directors

Jan L. Bergen

Cindy Bo, MBA | Secretary

Herman Bontrager | Chair of the Board

Elam Esh

Leon Hoover | Vice Chair

Leonard Hurst

Mark Martin | Treasurer and Chair-Finance Committee

Jacob Petersheim

Jonathan H. Salvin, MD

Stephen Tiftt, MD

Glen Zimmerman | Chair-Development Committee

Keeping the Promise:

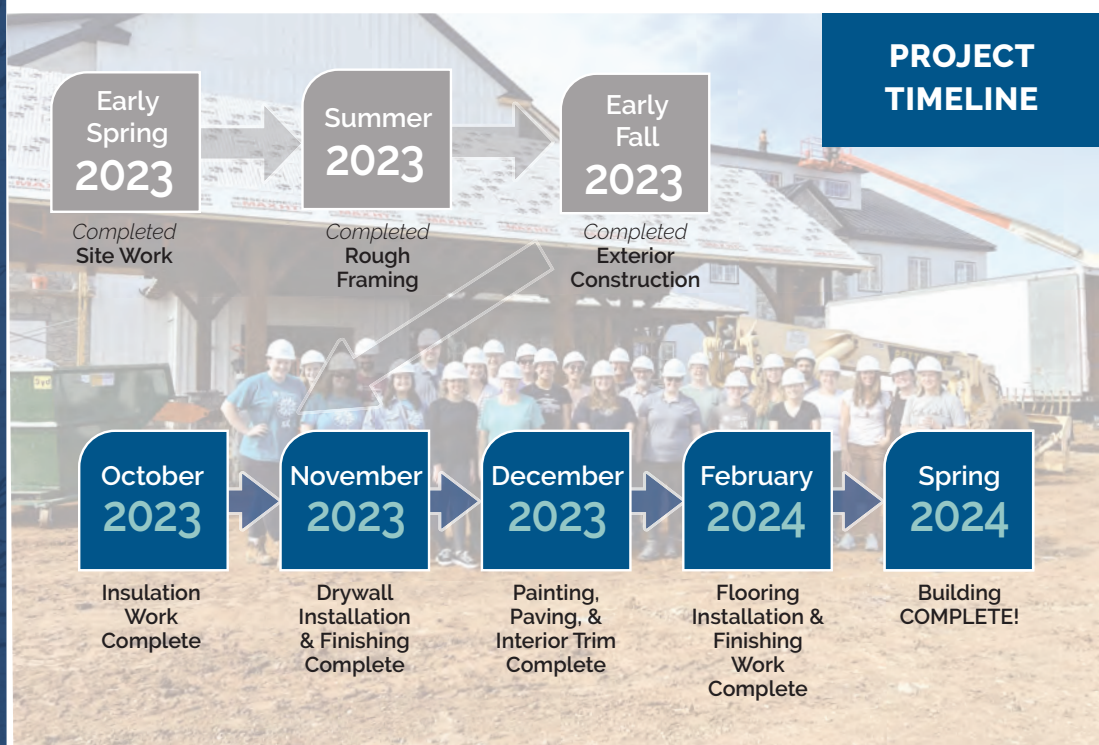
Our vision is becoming a reality. Since the last Clinic for Special Children newsletter in early summer, the new Clinic facility's exterior is largely complete! All roofing, exterior siding, and stonework installation is done. On the interior side, crews will be installing drywall, interior trim, and painting over the next few months.

On the fundraising side, we're eager to finish the campaign by March and we are currently at 79% of our \$12.5 million goal as of September 18, 2023. We're hoping to progress alongside of the construction process, so all funds are raised by the opening of the Clinic (currently planned for spring 2024). We greatly appreciate everyone that has supported the project, and we look forward to opening the doors of our new facility soon!

ON OUR WAY TO REACHING OUR GOAL

As of September 18, 2023
Over 2,180 supporters and
\$9.9 million raised (79% of \$12.5 million goal)

Funds Left to Raise
\$2.6 million





September 14, 2023
Finalizing exterior work



September 5, 2023
Nearing final stages of exterior work



August 30, 2023
The Clinic staff toured the new site!



August 24, 2023
Stonework & siding installation

Donations of In-Kind Materials and Labor

Many hands have made light(er) work! Thanks to many businesses and supporters, over \$1.6 million has been donated in gifts of in-kind labor and materials for our project.

Although we are entering the final stages of construction, there are still opportunities to help! We're still looking for volunteer labor and/or materials for periodic site cleaning/sweeping, painting, drywall installation, interior trim installation, and more.

If interested, please contact Elam Stoltzfus, Owner's Representative (info to the right). It's been inspiring to see so many individuals work together to build a new home for the Clinic.



Elam Stoltzfus
Owner's Representative
Narvon Builders
(717) 629-3159

STAY UP-TO-DATE!

Visit www.ClinicforSpecialChildren.org/campaign or call our project hotline at (717) 207-8607



**Clinic for
Special Children®**

535 Bunker Hill Road
PO Box 128
Strasburg, PA 17579

tel (717) 687-9407

fax (717) 687-9237

ClinicforSpecialChildren.org

NONPROFIT ORG.
U.S. POSTAGE
PAID
LANCASTER, PA
PERMIT NO. 1746

You can unsubscribe from our newsletter mailing list by calling us at 717-687-9407, emailing queries@clinicforspecialchildren.org, or mailing us (Clinic for Special Children, Attn: Kelly Cullen, PO Box 128, Strasburg, PA 17579).



The Clinic for Special Children is a Pennsylvania non-profit corporation and a 501(c)3 public charity for US federal and state tax purposes (Tax ID # 23-2555373). The official registration and financial information of The Clinic for Special Children, Inc. may be obtained from the Pennsylvania Department of State by calling toll free, within Pennsylvania, 1 (800) 732-0999. Registration does not imply endorsement.

2023 Extraordinary Give

Lancaster County's Largest Day of Online Giving!

Save the date for the 2023 Extraordinary Give on Friday, November 17th!
Details inside on how to help the Clinic fundraise during this 24-hour online giving marathon.

Our Vision is Becoming a Reality

Read inside to see the latest on our new building project!
We're excited to see our vision of the Clinic's future home becoming a reality.

The Clinic's Mission

"To serve children and adults who suffer from genetic and other complex medical disorders by providing comprehensive medical, laboratory, and consultative services, and by increasing and disseminating knowledge of science and medicine."

*Charles M. Hantrop D.D.
To 50 B.R. Harris 300 90*