Margherita Carboni – Indication Lead Neurology, Roche Diagnostics

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In conversation at Clinical Trials on Alzheimer's Disease (CTAD) 2024 in Madrid, Roche Diagnostics' Margherita Carboni discusses the potential of blood-based biomarkers to transform Alzheimer's diagnostics, offering a faster and less invasive alternative to traditional methods. She highlights Roche's commitment to ensuring diversity in clinical trials, a crucial step in developing tools that are effective for all patient populations. Additionally, Carboni sheds light on how Roche's integrated approach between its diagnostics and pharma divisions aims to streamline patient care from diagnosis to treatment.

How are patients generally diagnosed for Alzheimer's disease today?

Diagnosis usually begins when a patient or their family notices memory issues and seeks help, often at a primary or secondary care clinic. For years, many people assumed that symptoms of Alzheimer's were just part of aging or mistakenly equated it with general dementia. This misconception, among other issues, means that there are significant variations in care standards worldwide today. Initial diagnosis generally starts with cognitive assessments, which can be lengthy and involve answering detailed questions to evaluate memory and cognitive function.

At Roche, we've introduced CSF testing (using cerebral spinal fluid collected via lumbar puncture) to detect amyloid, the underlying pathology of Alzheimer's. This test, along with brain imaging,

provides a clearer diagnosis, combining cognitive and biomarker data to confirm if someone has Alzheimer's. However, this process is often long, can be costly, and is invasive. That's where blood-based biomarkers come in. We see immense potential in blood-based diagnostics, which could simplify and speed up the process with high accuracy, helping patients get answers sooner.

What are blood-based biomarker tests, and why might they represent a major shift in Alzheimer's diagnostics?

First, it's important to clarify that blood-based biomarkers are designed to detect amyloid pathology—one of the key markers of Alzheimer's disease. However, to diagnose Alzheimer's accurately, this information must be combined with other diagnostic data.

The advantages of blood-based biomarkers are clear: they're minimally invasive, relying on a simple blood draw. Currently, these tests are not yet in routine clinical use, but once validated and approved, they could significantly streamline the diagnostic process. At Roche, we're working on a system that makes this testing easily accessible. The collected blood can be quickly processed by machines that are already widely available, meaning this diagnostic tool could be accessible to a broad population, with fast turnaround times and minimal barriers.

What is the expected timeline for Roche's blood-based biomarker diagnostics to become available to patients?

Roche and other companies already have CSF tests on the market. Regarding blood-based biomarkers, Roche has received FDA Breakthrough Device Designations for both pTau181 and pTau217 assays—the former granted earlier, and the latter in 2024. We are conducting extensive clinical validations across diverse populations to ensure these tests are robust and reliable. We are working closely with regulators and anticipate that it could receive CE mark approval from late 2025 which could open up availability in countries accepting the CE mark. We work closely with all regulatory bodies across the world to make these diagnostics available globally as early as possible.

Can you tell us about the clinical trial data you're presenting here at CTAD, including the patient diversity and why that's so important in Alzheimer's research?

This trial is one of the largest of its kind, designed to reflect real-world practice as closely as possible. We intentionally kept our exclusion criteria minimal, aiming to include a broad spectrum of individuals. Our trial spans multiple geographies—Europe, the US, and Australia—with a strong focus on including racial minorities, especially within the U.S. This diversity is crucial, as Alzheimer's patients are often older and dealing with multiple health conditions. Ensuring a representative sample helps us create a diagnostic tool that is effective across different backgrounds.

During our interactions with the FDA, diversity was a central discussion point, and we're proud of the high proportion of minority participants in the study. This approach aligns with the industry-wide focus on ensuring that diagnostic products, once on the market, are effective and accessible for all patient demographics. The population we're studying will also be integral to our regulatory submissions, making this inclusivity both a scientific and regulatory priority.

Roche has both diagnostics and pharma divisions and is working on both Alzheimer's treatments as well as diagnostics. How do these two arms collaborate, and how does this impact patient care?

That's one of our unique strengths; having both the pharma and diagnostics divisions under one roof. We work hand-in-hand to create a seamless patient journey, from early diagnosis through to treatment and ongoing monitoring. This integrated approach ensures that patients benefit from a streamlined pathway, where diagnostics can guide treatment decisions, and treatments are followed up with precise monitoring.

Although we're one company, we maintain strict internal boundaries, or "firewalls," which allows our diagnostics team to work independently, including engaging in partnerships with other pharma companies. For instance, we at Roche Diagnostics collaborate with Eli Lilly, and we're open to partnerships with various pharma, biotech, and medical groups. We see collaboration as essential, especially in diagnostics, to advance medical education and development, ensuring that the benefits of our combined expertise reach as many patients as possible.

Being here at CTAD, there's a lot of excitement with new data readouts and discussions among peers. Beyond your own work, is there anything you've seen or heard that particularly excites you?

It’s hard to single anything out because there’s just so much happening! Of course, I’m especially excited about the data coming from our colleagues in Roche Pharma—it’s something the field really needs. Beyond that, I find the emergence of new biomarkers incredibly promising. Alzheimer’s is such a complex disease, and we’re now able to break it down into specific pathologies like amyloid and tau. It’s inspiring to see how new biomarkers are being developed to track these aspects more precisely.

Ultimately, the most important thing is keeping the patient at the center of all these advancements. Offering diagnostic solutions throughout the patient journey is crucial, and it’s encouraging to see the whole field moving in this direction.

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