

# Expanding NGS Data with Optical Genome Mapping (OGM)

NCT06851377

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<b>Status</b>	RECRUITING
<b>Phase</b>	Not Applicable
<b>Sponsor</b>	IRCCS Eugenio Medea
<b>Enrollment</b>	60 participants

## Key Eligibility Criteria

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### Inclusion (4)

- individuals without a molecular diagnosis (negative to ES/CMA analyses);
- individuals with genetic diagnoses that explain only one component of their primary phenotype;
- individuals carrying one or more variants of uncertain clinical significance
- individuals with a phenotype highly reminiscent of clinically and molecularly well-defined syndromes (i.e., Marfan Syndrome) but negative to routine molecular analysis.

### Exclusion (1)

- individuals who have not undergone initial diagnostic genetic tests (ES/CMA)

## Locations (1 total)

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Cytogenetic Unit of Medical Genetic Laboratory, Bosisio Parini, Lecco, Italy