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HUMAN GENETICS EXPERT OPINION

Social Security Expert Opinion | Occupational Disability / Reduced Earning Capacity

Marfan syndrome (MFS) with aortic and skeletal involvement | Occupational disability of a carpenter / craftsman | FBN1 pathogenicity confirmed

Case Number	SG [City] - Ref. No. S [XX] R [XXXX]/[YY] (Sample Case)
Client	Social Court [City] / German Pension Insurance [Region], Order for Evidence dated [Date]
Plaintiff	Mr. Thomas B. [fictitious], born March 15, 1978 (47 years old), male
Occupation	Carpenter / Master Carpenter; most recently self-employed; limited professional activity since [year], ceased work since [year]
Defendant	German Pension Insurance [Region] (denial of disability pension)
Expert	Prof. Dr. med. Christian T. Thiel, MBA
Qualifications	Board-certified specialist in human genetics; University Hospital Erlangen; 30 years of clinical and scientific experience; expert witness for courts and insurance carriers
Date	March 28, 2026
Diagnosis	Marfan syndrome (MFS; OMIM #154700); FBN1 mutation c.5788G>T (p.Gly1930Cys), ACMG Class 5; aortic root dilation (45 mm); bilateral lens dislocation; scoliosis 32 degrees Cobb
Key Issue	Is the plaintiff, due to confirmed Marfan syndrome with cardiovascular, ocular, and skeletal involvement, unable to work as a craftsman/carpenter, and does this constitute a complete loss of earning capacity within the meaning of § 43 SGB VI?

Declaration pursuant to § 407a ZPO

The undersigned declares that the expert opinion has been prepared impartially and to the best of his knowledge and belief. He affirms that he has no personal or financial relationship with the parties. The expert opinion is based on the documents submitted, his own examination, the current state of medical science, and the applicable principles of health care medicine (Health Care Medicine Ordinance, VersMedV).

1. Court's Mandate and Questions

The Social Court of [City] commissioned the undersigned by order for evidence dated [Date] to provide an expert assessment of the following questions:

1. Does the plaintiff, Mr. Thomas B., have Marfan syndrome confirmed by molecular genetic testing, and what clinical manifestations are documented?

2. What specific physical limitations result from Marfan syndrome and its complications (cardiovascular, ocular, skeletal) for the performance of manual labor?
3. Is the plaintiff still able to perform his most recent occupation as a carpenter/master carpenter on a full-time, part-time, or no longer at all basis?
4. Is there a total or partial loss of earning capacity within the meaning of Section 43 of Book VI of the Social Code (SGB VI)?
5. How should the degree of disability (GdB) be assessed according to the Principles of Medical Care (VersMedV)?
6. What activities could the plaintiff still perform on a full-time basis despite his illness (referred activities)?

2. Examination Materials and Methodology

2.1 Submitted documents

- Molecular genetic report: FBN1 sequence analysis (Date: [Date]); variant c.5788G>T (p.Gly1930Cys), pathogenic, ACMG Class 5
- Cardiological findings including echocardiography series [Date]-[Date]:
  Aortic root 45 mm, bicuspid aortic valve ruled out, no dissecting aneurysm
- Ophthalmological findings report: Bilateral lens dislocation, myopia -14 dpt. in both eyes, best-corrected visual acuity 0.4 / 0.3
- Orthopedic/spinal findings reports: Scoliosis 32 degrees (Cobb), lumbar disc protrusion L4/5, thoracic kyphosis
- Pulmonology Findings report: Spontaneous pneumothorax (Event [Date]), lung function FEV1 78%
- Pension decisions from the German Pension Insurance (Rejection [Date], Appeal decision [Date])
- Medical certificate and documentation from the treating cardiologist and orthopedist
- Workers' compensation accident file (fall while roofing [Date])

2.2 Our own examination

On [Date], Mr. Thomas B. was personally examined at the Institute of Human Genetics. The examination included: a clinical-genetic full-body examination with findings on dysmorphology, assessment of mobility and physical capacity, eye examination (orientative slit lamp), measurement and assessment of body proportions, and evaluation of all available external findings.

2.3 Legal Basis for the Assessment

The assessment of reduced earning capacity is conducted in accordance with Section 43 of SGB VI (Pension for Reduced Earning Capacity). Complete reduced earning capacity exists if the insured person is unable, due to illness or disability, to work for at least three hours daily under the usual conditions of the general labor market for an indefinite period. Partial reduced earning capacity exists when the remaining earning capacity is between three and less than six hours per day. The assessment of the GdB follows the Principles of Medical Care (VersMedV, Appendix 'Principles of Medical Care').

3. Medical History

3.1 Occupational history

After graduating from secondary school, Mr. Thomas B. trained as a carpenter and passed the master craftsman's examination in [year]. He worked as a salaried master carpenter from [year] to [year] and became self-employed in [year] (carpentry and roofing business, [X] employees). The work regularly involved:

- Heavy lifting and carrying (wooden beams, roof trusses; regularly > 25 kg, occasionally up to 80 kg)
- Working at great heights (roof trusses, scaffolding, up to 15 m high)
- Physically demanding forced postures (bending over, kneeling, overhead work)
- Exposure to vibration (chainsaw, drill, nail gun)
- Exposure to weather conditions (cold, wet, heat)

3.2 Medical History

The diagnosis of Marfan syndrome was first made in [Year] at the age of [Age] after the plaintiff was hospitalized for a spontaneous pneumothorax. The clinical diagnosis was confirmed in [Year] by molecular genetic detection of the FBN1 mutation. The following complications were documented over time:

- [Year]: Spontaneous pneumothorax on the left, treated with chest drainage; later on the right (recurrent)
- [Year]: Initial echocardiography: aortic root 38 mm; annual follow-ups since then; currently 45 mm
- [Year]: Ophthalmological diagnosis of bilateral lens dislocation; surgical correction on the left, conservative management on the right
- [Year]: Fall from scaffolding (from approx. 3 m in height) with rib fracture on the right; covered by workers' compensation
- [Year]: Discontinuation of physical activities activities due a cardiologist's recommendation due to a 43 mm aortic root
- [Year]: Cessation of employment; application for disability pension; denied by the German Pension Insurance (DRV)

3.3 Current symptoms (at the time of examination)

- Exertional dyspnea starting with mild physical exertion (NYHA II-III)
- Back pain with prolonged standing and sitting (scoliosis, disc protrusion)
- Visual impairment: best-corrected visual acuity 0.4 / 0.3; no longer eligible for a driver's license
- Joint pain and instability in both knees and both hands (hypermobility)
- Dizziness and balance disorders (partially due to ocular causes)
- Psychological stress due to chronic illness and anxiety about the future (documented adjustment disorder)

4. Family history and hereditary factors

Marfan syndrome is inherited in an autosomal dominant pattern with very high penetrance. Approximately 75% of cases are familial, while about 25% result from de novo mutations.

- Father (deceased at age 52): Type A aortic dissection; retrospectively consistent with Marfan syndrome, not confirmed by molecular genetic testing
- Brother (44 years): clinically suspicious (tall stature, Marfanoid appearance); genetic counseling recommended, not yet performed
- Plaintiff's son (18 years old): no symptoms to date. Predictive testing for FBN1 mutation recommended

The family history (particularly the father's sudden cardiac death) underscores the clinical relevance of cardiovascular involvement and confirms the autosomal dominant pattern of inheritance.

5. Clinical findings

5.1 Anthropometric findings / proportions

Auxiology: Height 194 cm, wingspan 204 cm (wingspan/height ratio 1.05; normal < 1.03), upper/lower body length ratio 0.86 (normal > 0.89). Arachnodactyly: Walker-Murdoch sign positive bilaterally, Steinberg sign positive bilaterally.

5.2 Cardiovascular findings (based on current echocardiography)

- Aortic root (sinus valsalvae): 45 mm (Z-score +3.8; age- and height-adjusted normal value < 37 mm)
- Ascending aorta: 38 mm (suspected dilation, borderline range)
- Mitral valve: Mitral valve prolapse, mild mitral regurgitation, Grade I
- No aortic dissection, no pericardial effusion
- Cardiology recommendation: Avoidance of physically strenuous activities; long-term beta-blocker therapy; echocardiographic follow-up every 6 months; surgical indication if > 50 mm or rapid progression

5.3 Ophthalmological findings

- Ectopia lentis (lens dislocation) bilaterally: surgically treated on the left, conservative management on the right
- High myopia: -14 dpt. left, -12 dpt. right
- Best-corrected distance visual acuity: 0.3 in the right eye / 0.4 in the left eye (significant visual impairment)
- No glaucoma, no retinal detachment at present; increased risk of retinal detachment due to myopia

5.4 Skeletal findings

- Scoliosis: thoracolumbar, Cobb angle 32 degrees, right-convex
- Thoracic kyphosis: 55 degrees (normal < 45 degrees)
- Lumbar disc protrusion at L4/5 with nerve root irritation (confirmed by imaging)
- Generalized joint hypermobility (Beighton score 7/9)
- Flat feet on both sides; pes planovalgus

5.5 Pulmonary findings

- Condition following two spontaneous pneumothoraces (left, right)
- Lung function: FEV1 78 % Target,FVC 82 % Target;mild restrictive ventilatory disorder
- Bullae and areas of emphysema bilaterally in the apical regions (confirmed by CT)

6. Molecular genetic findings

6.1 FBN1 variant

Molecular genetic findings - FBN1 sequence analysis Gene:
FBN1 (Fibrillin-1; chromosome 15q21.1; NM_000138.5) Variant:
c.5788G>T (p.Gly1930Cys) - heterozygous
Variant type: Missense; glycine-to-cysteine substitution in an EGF-like repeat of the fibrillin-1 domain
Classification: Pathogenic (ACMG Class 5)
Reference: OMIM #154700 | Ghent Nosology 2010: fully met

6.2 Pathomechanism and clinical significance

Marfan syndrome is caused by pathogenic variants in the FBN1 gene, which encodes the structural protein fibrillin-1. Fibrillin-1 is an essential component of the extracellular microfibrillar networks that surround elastin and are responsible for the mechanical stability of connective tissue —particularly in the aortic wall, the zonular fibers of the eye, and the skeleton. [1,2]

The identified glycine substitution p.Gly1930Cys is a characteristic type of FBN1 missense variant in which the glycine residue is replaced in a calcium-binding EGF-like repeat. **This class of FBN1 variants is considered to be particularly strongly associated with cardiovascular involvement (aortic dilatation, aortic dissection).** [3,4] The diagnosis according to the 2010 Ghent Nosology [2] is fully met based on the combination of aortic dilatation (Z-score > 2), lens dislocation, skeletal involvement (system score > 7), and a pathogenic FBN1 variant.

7. Genetic and clinical evaluation

7.1 Diagnosis and completeness of clinical presentation

The diagnosis of Marfan syndrome is molecularly confirmed in the plaintiff and is clinically fully manifested. The complex of findings (aortic root 45 mm, bilateral lens dislocation, scoliosis 32 degrees, spontaneous pneumothorax, Marfanoid body proportions) is characteristic and, in its combination, unambiguous from a human genetic perspective. There is no diagnostic doubt.

7.2 Risk profile and limitations due to cardiovascular involvement

The aortic root dilation of 45 mm represents the most life-threatening manifestation. **Starting at an aortic root diameter of 45–50 mm, there is a significant risk of aortic dissection, which is fatal in up to 90% of cases if not treated surgically immediately. [1,5]**

The following is crucial for assessing occupational disability: Any physical exertion that could increase blood pressure and aortic wall tension (isometric exercises, heavy lifting, straining, risk of falling) is contraindicated from a cardiological and human genetic perspective when the aortic diameter is 45 mm. The current guidelines of the European Society of Cardiology (ESC 2022) [5] list the following absolute contraindications:

- Lifting loads > 5–10 kg
- Isometric physical exertion (lifting, pressing)
- Work involving a risk of falling or an increased risk of injury
- Jerking movements, exposure to vibration
- Working under time pressure and psychological stress (increased blood pressure)

All of these contraindications are central components of a carpenter's professional activities.

8. Assessment of occupational disability and reduced earning capacity

8.1 Occupational disability as a carpenter / master carpenter

According to the job description and relevant occupational sources, working as a carpenter/master carpenter regularly entails the following physical requirements:

Occupational requirement	Reasonable for the plaintiff?	Reason
Lifting/carrying > 10 kg	**NO**	Contraindicated with Ao root 45 mm (ESC guideline)
Working at heights (scaffolding, roof)	**NO**	Risk of falling; consequences of a fall can be potentially fatal in cases of aortic aneurysm; visual impairment (visual acuity 0.3/0.4) increases the risk of falling
Exposure to vibration (chainsaw, nail gun)	**NO**	Vibration increases aortic wall tension; contraindicated in cases of aneurysm
Heavy physical labor (> 6 hours)	**NO**	Exertional dyspnea NYHA II–III; cardiopulmonary impairment
Work in forced positions (bending over, kneeling)	**NO**	Scoliosis, L4/5 disc herniation, joint hypermobility; unable to perform due to pain
Fine manual work (seated, light)	Limited	Possible for max. 2–3 hours/day; limited by visual impairment
Office work / administrative management tasks	Limited	Possible, but visual impairment limits computer work; retraining in a different field is necessary

Conclusion: The plaintiff is no longer able to work as a carpenter or master carpenter—not even on a reduced schedule**. Occupational disability within the meaning of insurance law exists.**

8.2 Reduced earning capacity pursuant to § 43 SGB VI

To determine eligibility for a reduced earning capacity pension, it must be assessed whether the plaintiff can still perform any work on the general labor market on a full-time basis (> 6 h/d) or part-time basis (3–6 h/d):

- Physically moderate to heavy activities: Completely excluded (aortic findings, cardiopulmonary impairment)
- Light physical activities while standing or involving frequent changes in posture: max. 2–3 hours/day possible (back pain, joint hypermobility, fatigue)
- Computer work / administrative tasks: significantly limited due to visual impairment (best-corrected visual acuity 0.3/0.4); tolerable for a maximum of 2–3 hours per day
- Precision work under time pressure: significantly limited due to visual impairment and tremor (side effect of beta-blockers)
  side effect)
- Work on ladders, scaffolding, with vehicles: completely excluded (visual impairment falls below the threshold for driving eligibility)

Overall assessment: The plaintiff's remaining capacity is less than 3 hours per day for any activity in the general labor market. **There is a complete loss of earning capacity within the meaning of Section 43(2) of SGB VI.**

8.3 Degree of disability (GdB)

According to the Principles of Medical Care (VersMedV, Annex), the GdB is to be determined based on the individual GdBs of the functional limitations (no simple addition; overall assessment):

Functional limitation	Individual GdB (estimate)	Basis: VersMedV
Aortic root dilation 45 mm, mitral valve insufficiency Grade I	40–50	Cardiovascular diseases
Bilateral lens dislocation, visual acuity 0.3/0.4	30–40	Eyes (visual impairment in both eyes)
Scoliosis 32 degrees + L4/5 disc herniation + thoracic kyphosis	20–30	Spine / Skeleton
Spontaneous pneumothorax on both sides, mild ventilation impairment	10–20	Lungs
Joint hypermobility with functional impairment	10	Extremities
Adjustment disorder / psychiatric comorbidity	20–30	Mental health
Total GdB (overall assessment)	80–90	No addition; overall assessment

Recommendation: GdB 80, possibly 90 depending on the progression of the aortic condition. Special codes G (significant walking impairment due to scoliosis and cardiopulmonary limitations) and B (requirement for an accompanying person on public transportation due to visual impairment and tendency toward dizziness) should be considered.

9. Conclusion / Expert Assessment

Summary of the expert findings

1. Confirmed diagnosis: The plaintiff has a molecularly and clinically confirmed Marfan syndrome (OMIM #154700), caused by the pathogenic FBN1 variant c.5788G>T (p.Gly1930Cys, ACMG Class 5). The Ghent Nosology 2010 criteria are fully met.
2. Multisystemic manifestations: The condition manifests in the plaintiff as cardiovascular (aortic root 45 mm, mitral valve prolapse), ocular (bilateral lens dislocation, visual acuity 0.3/0.4), skeletal (scoliosis 32 degrees, disc protrusion, hypermobility), and pulmonary (condition following spontaneous pneumothorax, mild ventilatory impairment).
3. Inability to work as a carpenter: The plaintiff is no longer able to perform his most recent occupation as a carpenter/master carpenter for any relevant number of hours. The typical occupational requirements (lifting, working at heights, exposure to vibration, heavy physical labor) are absolutely contraindicated due to the cardiovascular and ocular involvement.
4. Total disability: The plaintiff's residual capacity is limited to less than 3 hours per day for any activity in the general labor market. Total disability within the meaning of Section 43(2) of SGB VI is present.
5. GdB and disability codes: A GdB of 80 (or 90 if applicable) is recommended. Assessment of disability codes G and B is indicated.
6. Permanence: Marfan syndrome is a lifelong, genetically determined condition. The limitations are likely to increase over time (aortic progression, ocular complications). A significant improvement in earning capacity is not to be expected.

The above expert findings are rendered to the best of my knowledge and belief, impartially and conscientiously, in accordance with § 407a ZPO. The undersigned certifies that he is not in a relationship of dependency with any of the parties.

9a. Referral Activities and Rehabilitation Potential

The court must determine whether a referral activity is reasonable for the plaintiff. From a clinical human genetics perspective, the following activities may still be considered for the plaintiff, but only for a limited number of hours (< 3 h/d):

– Consulting work in the construction industry (sedentary, without physical strain) – however, this requires retraining and presupposes sufficient visual acuity

- Teaching theoretical content in carpentry training (vocational school) – subject to the limitation of visual impairment for blackboard and screen work
- Administrative office work—limited to a maximum of 2–3 hours per day due to visual impairment

None of these activities can reasonably be expected of the plaintiff on a full-time or even part-time basis (> 3 hours per day). **No referral activity that meets the requirements of Section 43 of SGB VI is apparent.**

9b. Medical-Social Assessment and Recommendations

- Pension insurance: Application for full disability pension under § 43(2) SGB VI is justified; approval recommended
- Disability Law: Apply for GdB 80; compensatory measures (protection against dismissal, tax exemption, increased travel expense reimbursement) apply for GdB >= 50
- Long-term care insurance: No care level based on current findings; re-evaluate following aortic surgery or further progression
- Workers' Compensation Association: Fall accident ([year]) is a Workers' Compensation case; long-term consequences should be reviewed for a Workers' Compensation pension
- Rehabilitation: Medical rehabilitation (cardiological and orthopedic) is advisable, but not with the goal of restoring the ability to work as a carpenter—this goal is realistically unattainable
- Psychosocial support: Psychooncological/psychosomatic support is strongly recommended for existing adjustment disorder

10. References

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